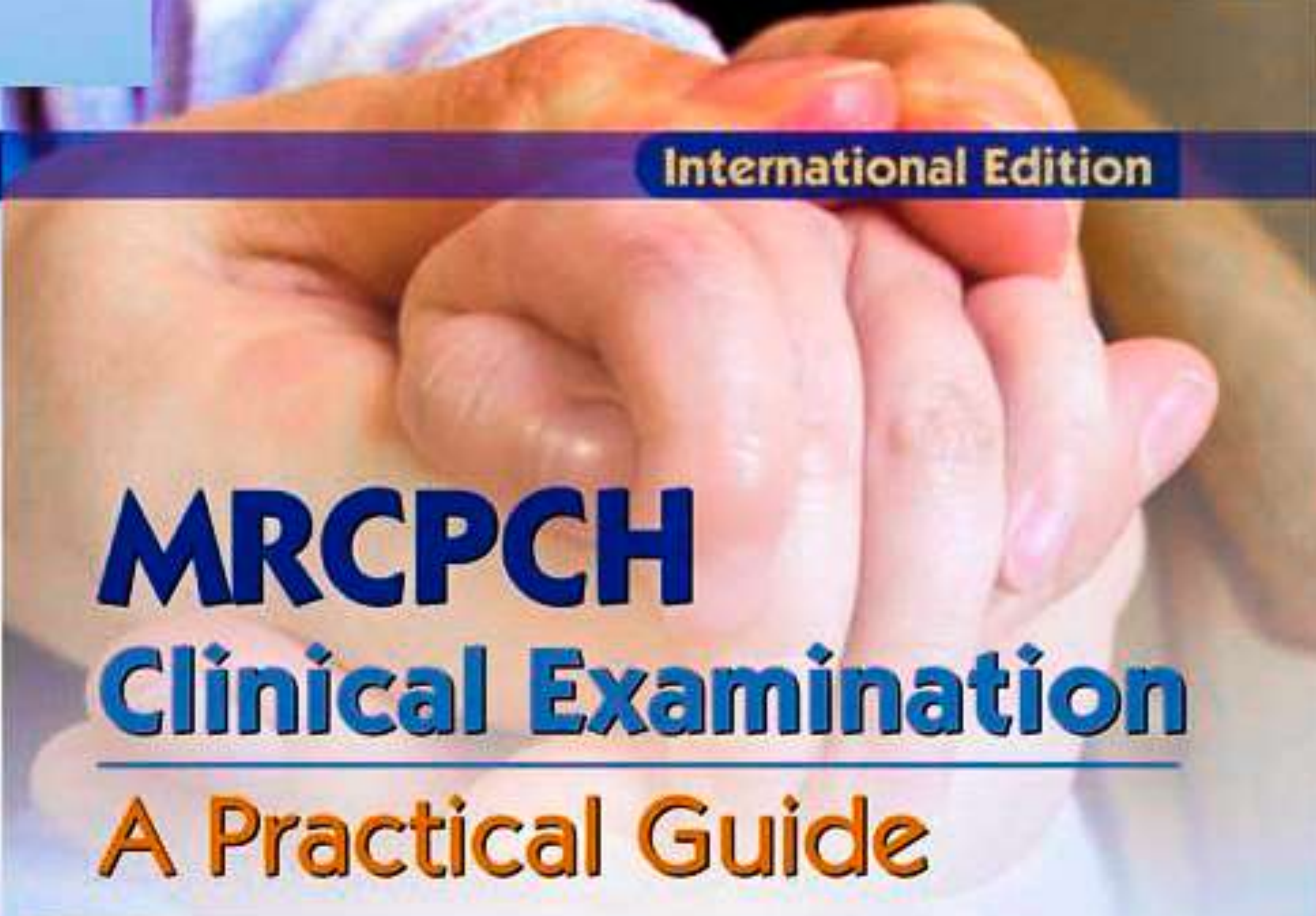

International Edition

MRCPCH Clinical Examination

A Practical Guide

Dr. Abraam Hanna

Second edition

مقدمة

زملائي الأعزاء، أقدم لكم هذا الكتاب مجاناً، وهو خلاصة خبرة 4 سنين من الاعداد ل امتحان زمالة الكلية الملكية لطب الأطفال الجزء الثالث.

أقدمه لكم ظناً مني انه فعلا كتاب قيم يحتوي علي Approaches منظمة و Additional notes قيمة و differential diagnoses كاملة ، كل ذلك كفيل بان يعطيك مساعدة قوية للاستعداد للامتحان.

مراجع الكتاب:-

= كتاب مارك بيتي 2007
= كتاب د. مصطفي البابا 2009
= محاضرات د. معتز 2015
= مذكرات د. شريف دبور 2016
= محاضرات د. خالد كمال 2016
= محاضرات د. محمد نصر و د. رضا 2018/2017

تعمدت أن أكتب هذه المقدمة بالعربية ، لأني أقدم الكتاب لزملائي المصريين و جميع المتحديث باللغة العربية ، سائلاً المولي عز و جل ان يستخدم هذا المجهود البسيط لمساعدة الزملاء ف اجتياز هذا الامتحان.

بالتوفيق للجميع
Abraam H. 2018

CARDIOLOGY

General observation:- well, not in distress, built for age **+ cyanosis** + surroundings **+ Growth** + dysmorphic features

Examination:- **must be in 45 degrees**

Hand:-

- Clubbing
- Cyanosis
- Pallor
- Stigmata of endocarditis
- Pulse (first count on one side then check bilateral for equality)
  Rate, rhythm, volume, equality on both sides
  (if you are not sure about the pulse or weak or not detected….>> please do not talk at all, but just keep in mind AS ?? and wait till the end)
- WATER HAMMER PULSE

Cases

- VSD
- PS
- AS
- AR
- MR
- TOF
- COMPLEX CYANO
- PDA

Head:-

- Conjunctiva (pallor / Jaundice : if you suspect Alagille)
- Stick your tongue out (cyanosis)
- Teeth

Syndromes

- Down
- Turner
- Noonan
- Williams
- Marfan

Neck:-

- Palpate carotids for thrill

Inspection :- **: Expose the chest and inspect for five things:**

- Scars
- Precordial pulge (feet and side of the patient)
- Visible pulsations
- Dilated veins

palpation:-

- Apex
- Thrills: upper lt sternal>>>PS
  Upper Rt sterna>>>AS

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Lower Lt sternal>>>>VSD
Suprasternal>>>>>>AS

Auscultation:-

- FRONT :- 4 areas + Axilla
- 1st and 2nd heart sound
- MURMUR
    - Apex >>>>mitral>>>if + >>>>>Axilla
    - Lower Lt sternal border>>>>>Tricuspid, VSD, AVSD
    - Lt 2nd intercostal>>>>>>>>>>>Pulmonary, PDA
    - Rt 2nd intercostals>>>>>>>>>>Aortic
  - RADIATION
    - To neck>>>carotid
    - To back>>PS (between scapulae)
    - To Axilla>>Mitral
    - To tip of the Lt scapula>> coarctation
    - To most of the chest wall>> VSD (not to the back)
  - Comment on a murmur :
    - Timing, grade, max. heard at, radiated to, and changing with position **for example:-** harsh pan systolic murmur, grade 3, heard all over the precordium with maximum intensity over the apex, radiated to the axilla, not changing with repositioning.

Back :- "HUG your SELF"

- Scars
- Scoliosis
- Auscultate :- interscapular + lung bases for crepitations

ABDOMEN:- ask for permission

- **Inspection:-** scars of PD post bypass surgery.
- **Palpation / percussion:-** hepatomegaly, liver span, ascites … splenomegaly ??

Lower limbs:-

- Clubbing
- Edema

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In order to complete:-

- BP
- JVP
- Peripheral pulses including femorals for Radio-femoral delay
- Examine the groin for any catheter or central lines scars
- O2 saturation

Management:-

- Thorough HX
- INVX:- X-ray, ECG, ECHO
    - In cyanotic pt :- FBC, HCT, Coagulation profile

Advice :-

- Keep good dental hygiene
- Avoid tattooing and piercing
- No exercise restriction except in AS

ABX prophylaxis:- Not advised in general except in case of Genitourinary or GIT procedures when there is expected infection at the surgery site after advice of the surgeon

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Cases

VSD:-

Signs of large VSD:-

- ✓ affecting growth
- ✓ in failure
- ✓ soft murmur
- ✓ loud P2

TOF:-

- Blue:-
    - Cyanosed, clubbed
    - Lateral thoracotomy scar
    - Shunt murmur

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    - PS murmur
- Pink:-
    - May be clubbed or not
    - Median sternotomy scar
    - May be lateral thoracotomy scar

Medical associations:- strokes, brain abscesses

Advice :- Hydration

Pulmonary stenosis:-

- Make sure that the pt is not Noonan
- Treatment options:- to be treated when pressure gradient>60
    - Baloon valvuloplasty
    - Surgical replacement when :-
        - Failure of balloon
        - Fixed subvalvular stenosis
        - Thickened valve like in Turner syndrome

Aortic stenosis:-

- Make sure that the pt is not Williams or Turner
- Ask the pt to bend forward while Auscultating
- Advice:- restriction of exercise
- TTT:- Balloon / Surgical replacement

Aortic Regurgitation:-

- Make sure that the pt is not Marfan or Ehler's Danlos
- Water hammer pulse
- Characteristic murmur
- Ask the pt to lean forward for Auscultation
- I want to look for peripheral signs of AR :-
    - pistol shot sign → over the brachial aretry
    - Duroziez's sign →Over the Femoral artery
    - corrigan's sign → visible pulsations over the carotids at the base of the neck
- Surgery:- if there are exercise intolerance symptoms

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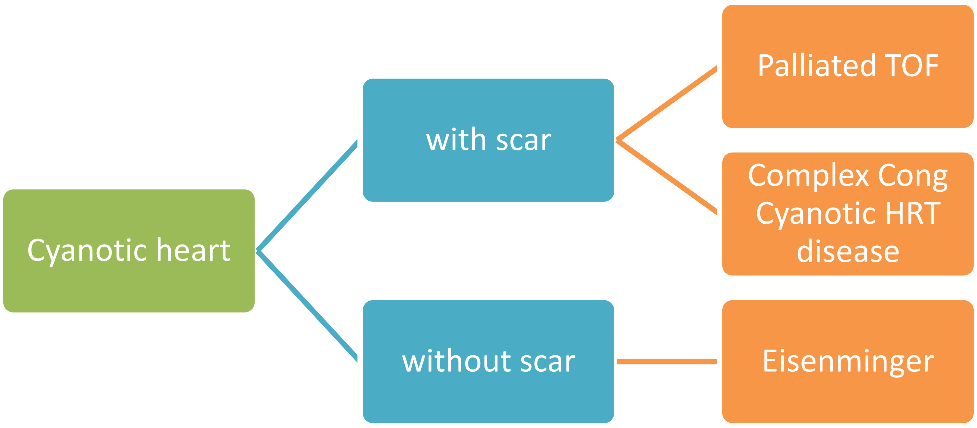
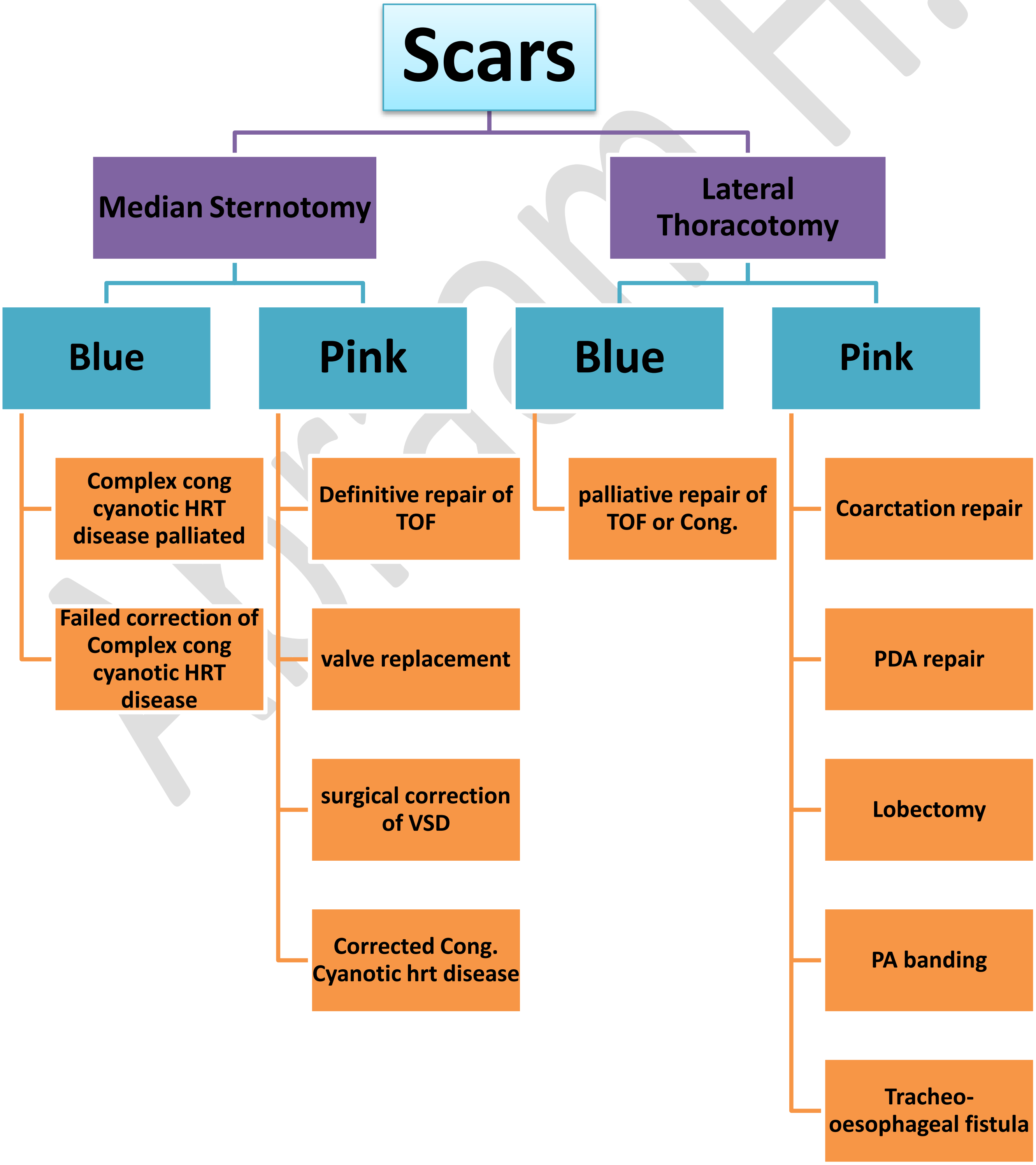


- Cyanotic heart
  - with scar
    - Palliated TOF
    - Complex Cong Cyanotic HRT disease
  - without scar
    - Eisenminger



- Scars
  - Median Sternotomy
    - Blue
      - Complex cong cyanotic HRT disease palliated
      - Failed correction of Complex cong cyanotic HRT disease
    - Pink
      - Definitive repair of TOF
      - valve replacement
      - surgical correction of VSD
      - Corrected Cong. Cyanotic hrt disease
  - Lateral Thoracotomy
    - Blue
      - palliative repair of TOF or Cong.
    - Pink
      - Coarctation repair
      - PDA repair
      - Lobectomy
      - PA banding
      - Tracheo-oesophageal fistula

Ahlam H.

RESPIRATORY

General observation:- as usual + cyanosis + distress + surroundings + Sputum Pot, coughing, snoring, stridor, attached devices, nutritional status, dysmorphism

Examination:-

Hand:-

- Clubbing
- Cyanosis
- Pallor
- Tremors
- Pulse

Cases

- Asthma
- Cystic fibrosis
- Bronchiectasis
- Kartagner
- CP + Pneumonia

Head/Neck:-

- Conjunctiva (pallor)(Jaundice in CF)
- Stick your tongue out (cyanosis)
- Teeth
- Lymph nodes (ممكن ماتعملوش بس اذكره ف الآخر)
- Trachea (ask for permission from the examiner)

Front →EXPOSE THE CHEST (look at the abdomen for a clue if CF)

inspection:-

- **RR**
- من عند الرجلين asymmetry, movement
- من الجنب Antro posterior diameter (^ in asthma)
- عادي Scars,Shape (normal, Barrel) Deformity (pectus carinatum, excavatum, Harrison's sulci)

Palpation:-

- Apex
- Chest expansion
- Tactile vocal fremitus (44)

Percussion:-

- Apex of the lung + clavicle + upper + middle + lower + liver span

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Auscultation:- **For air entry, breath sounds and additional sounds**

- Upper + middle + axilla + lower

Back →Hug yourself (لازم اقعد ع السرير ورا العيان)

inspection:-

- Skin (May be café au lait)+ Scars + Kyphosis + scoliosis

Palpation:-

- Chest expansion
- TVF

Percussion:-

- Suprascapular + paravertebral + subscapular

Auscultation:-

- Suprascapular + paravertebral + subscapular

Abdomen :- full abdominal examination (especially in cases of CF)

- **Inspection:-** Scars of meconium ileus or DIOS, Gastrostomy, Caput medusae
- **Palpation:-** Hepatosplenomegaly
- **Percussion:-** Ascites

In order to complete :-

- Lymphnodes
- ENT
- BP
- BCG scar
- Sputum put
- spirometry
- O2 Saturation
- Full CVS examination to look for signs of congestive heart failure secondary to PHTN (raised JVP, dependent edema, hepatomegaly)

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علق ع ال   مكانها و زمانها

Crackles :- fine or coarse , it's timing

Your management?

- **In asthma:**

1. I have to take a thorough history about his condition , compliance on medications, inhalation techniques, counseling about preventers(Asthma)
2. I would like to do some investigations
   CBC, PEFR, peak flow diary (diurnal variation), PFT (> 5 years), oxygen saturation, x ray chest.
3. MDT involvement in the form of the pulmonologist, my consultant, dietician, school.

- **In CF:**
  1. Through family history, genetic counseling, neonatal history, surgical history, diet (high calorie high protein) medications(pancreatic enzyme supplements, vitamins, antibiotics), physiotherapy and compliance.
  2. Hospital admission , saturation monitoring, Sweat test, blood cultures, sputum culture, inflammatory markers, immunoglobulins, peak flow, PFT, CBC, x ray chest
  3. (**CF TEAM**) in the form of pulmonologist, my consultant, phisotherapist, gastroenterologist and hepatologist, endocrinologist, dietician, psychologist and school, CF nurse.

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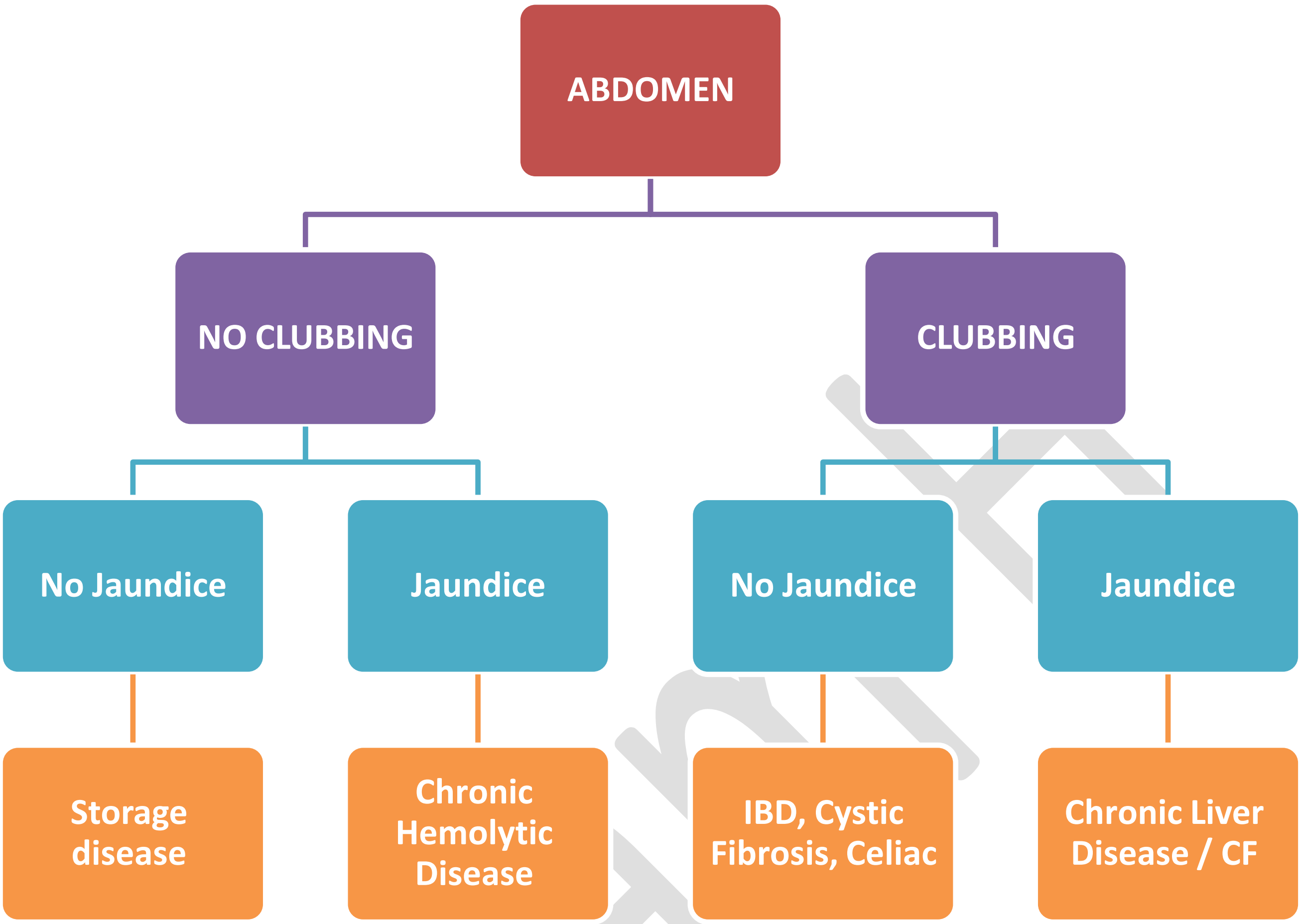
ABDOMEN


ABDOMEN
NO CLUBBING
CLUBBING
No Jaundice
Jaundice
No Jaundice
Jaundice
Storage disease
Chronic Hemolytic Disease
IBD, Cystic Fibrosis, Celiac
Chronic Liver Disease / CF

Other cases:- **HEPATOMEGALLY FOR DD, HSM FOR DD**

General Observation:- usual +

Examination:-

Hand:-

- CLUBBING
- Cyanosis
- Pallor
- Puncture sites
- Liver failure stigmata (palmer erythema , flabbing tremors)
- Pulse
- look to the forearm may be a site for heomodialysis
- Deltoid → sites of injection of desferral

Head:-

- Dysmorphology
- Eye:- (JAUNDICE)
- Mouth:- (اقلب الشفايف)dental caries(reflux), tongue ulcers or pigmentations

Neck:-

- LN (Virchow)
- Spider Naevi

EXPOSE CHEST AND ABDOMEN

CHEST:-

- Gynecomastia
- spider naevei
- subclavian scar (site for hemodialysis)
- Hickman line

Inspection Abdomen:- Ideally exposure is from the knee to the nipple line (شوف شمال و يمين)

- Shape / contour / distension
- Scars (Measure)
- Dilated veins
- Umbilicus shape and position
- Hypo or hyperpigmentations(thalassemia… desferral)

Palpation Abdomen:- دفي ايدك، اسأله لو فيه حاجة بتوجعه، اتكلم معاه، بص عليه علشان ال Tenderness

- Superficial for tenderness & rigidity
- Deep for masses (anti clock wise)
- Deep for organs:-
    - **Liver:-** (**tenderness**, size below costal margin, span, surface <**smooth**/nodular> , border < sharp/**rounded**>, Consistency < **firm**/hard>) It lobe below the Xiphisternum.
    - **Spleen:-** (same + Notch, move with respiration, can't get above it)
    - **Kidneys:-** (Ballotable)

Percussion :-

- From Xiphisternum till symphysis pubis (for any midline mass)
- For ascitis → Shifting dullness if there is ascitis
- Liver span
- To differentiate between SPLEEN (dull) & KIDNEY (resonant)

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Auscultation:-

- Bowel sounds
- Renal angles for renal artery stenosis
- Bruit of enlarged liver or suspected renal problem
- A-V fistula if present

BACK:- **Divarication of Recti**
Scars, scoliosis, auscultate renal angles

LOWER LIMBS:- for (erythema nodosum, oedema, ulcers in sickle cell)

In order to complete:-

- Hernial orifices
- Anal and rectal examination
- Tanner staging of puberty
- Lymph nodes examination

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Thalassemia

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Facial features:-

- ✓ Frontal bossing
- ✓ Maxillary hyperplasia
- ✓ Mandibular prognathism
- ✓ Dental crowdening

Investigations:

CBC+ Blood film (microcytic hypochromic anemia, target cells, basophilic stippling and nucleated RBC), Ret% + bilirubin, Liver function

Hb electrophoresis(HbF 70%-90%, HbA 10-20%, HbA2 2%).

MANAGEMENT:

- **Transfusions>>** 4-6 weekly, First time when Hb <6 g\dl

Then>> transfuse when Hb < 10 g/dl (hypertransfusion regimen).

- **Iron chelation:-** → start if ferritin level > 1000ug\L
    - SC Desferral(HMW… not oral) for 8-12 hours overnight 5 nights/week>> monitor ear , eye, Renal F. and bone..pseudorickets).
    - Defriprone → arthralgia, neutropena
    - Desferosirox → skin rash, ear & eye complications
- **Monitoring and TTT of complications of hemosiderosis:-**
    - Hypogonadism,
    - hypothyroidis,
    - short stature,
    - DM,
    - cardiomyopathy,
    - liver cell failures
    - Measure serum ferritin (1000-2000ug/L)
- **Folic acid 5 mg/day**
- **Splenectomy if:-**
    - Age > 5 years
    - requirment increased > 180- 250 ml /kg /year PRBCs
    - HUGE SPLEEN>>causing discomfort + increased risk of traumatic rupture
- **BMT**

COMPLICATIONS:

1. **Heamosiderosis:-** heart failure, arrhythmia (cardiomyopathy), liver cirrhosis. Hypothyroidism, Hypoparathyroidism, DM, hypopuituitarism & delay puberty.
2. **Iron chelation therapy side effects**
3. **Hazards of blood transfusion**

In splenectomy:- Vaccination:- pneumococal -meningeococcal -haemophilus plus + prophylactic penicillin

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Renal transplant cases

- ✓ Scar at the renal angle
- ✓ Scars of previous hemodialysis and peritoneal dialysis

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MANAGEMENT:

- I would like to take a detailed history, his medications
- **Investigations:** CBC, RFT, ABG, GFR, SERUM FERRITIN, PROTEIN AND ALBUMIN, CA, PO4, ALP AND X-RAY LEFT WRIST, MONITOR B/P AND URINE
- **MDT in the form of :-** My consultant =Nephrologist =Surgeon =dietician = immunologist =psychologist =support group =health visitor =school

COMPLICATIONS OF KIDNEY TRANSPLANT:

.Rejection .infection .recurrence (autoimmune) .leaking . Thrombosis

. post transplant lymphoreticular disease . Side effects of steroids

. Side effects of heamodyalisis:- hypotention and electrolytes disturbance**.**

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Hereditary Spherocytosis

- ✓ Mild pallor
- ✓ Mild jaundice
- ✓ Splenectomy scar (or splenomegaly)
- ✓ +- Kocher scar
- ✓ No liver

INVX:- FBC, BF, retics, Osmotic fragility test

Advice:- about vaccines and penicillin for splenectomy

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Sickle cell disease

- ✓ Mild jaundice
- ✓ Mild pallor
- ✓ +- spleen
- ✓ +- splenectomy scar
- ✓ +- liver
- ✓ +- Kocher scar

In order to complete my examination (look for associations):-

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- Neurological examination → Hemiplegia
- MSK and hip exam →Avascular necrosis of the femoral head
- Vision examination → eye affection by thromboemboli (proliferative retinopathy)
- Limb examination → dactylitis and ulceration

Sequestration crisis:-

- Sudden enlargement of the spleen
- Decreased HB by > 2gm%
- High retics
- Hypovoleamic shock

ttt →fluids, blood transfusion and splenectomy

Treatment of crises :- O2, analgesia, hydration, ABX, + blood transfusion and hydroxyurea

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Storage diseases

Glycogen storage

- ✓ Short stature
- ✓ Doll facies
- ✓ No pallor
- ✓ No jaundice
- ✓ Very huge hepatomegaly
- ✓ +- Mild splenomegaly
- ✓ May have some bruises

INVX:- FBC, BF, retics, LFT, UE, serum Glucose, Uric acid, Lactate, PLT function & liver biopsy & enzyme assay

Advice:- avoiding hypoglycemia by overnight feeding , glucose infusion, corn starch

Definitive ttt:- liver transplant

To differentiate between type 1 and 3 :-give glucose and measure lactate (in 1 decreased and in 3 increased).

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Hepatosplenomegaly

Don’t forget to comment on short stature and other features

Gausher

Huge splenomegaly + mild hepatomegaly , not pale , not jaundiced

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CAUSES OF HEPATOMEGALLY:

1. Infection (hepatitis A,B, EBV)
2. Infiltration>>GSD and other storage diseases(Gaucher, NP)
3. Tumors
4. OBSTRUCTIVE>>Right sided heart failure and hepatic vein thrombosis.
5. Idiopathic
6. Miscellaneous >> Wilson, alpha 1 antitrypsin deficiency, congenital hepatic fibrosis.

CAUSES OF SPLENOMEGALLY:

1. PORTAL HYPERTENTION
   a. Pre-hepatic:- Portal vein thrombosis
   b. Inra-Hepatic
      - Pre-sinusoidal
         - Neopasia, schistomiasis, hepatic cyst
      - Sinusoidal
         - Congenital hepatic fibrosis, cirrhosis, biliary atresia, neonatal hepatitis, Alpha 1 antitrypsin deficiency
      - Post-sinusoidal
   c. Post- hepatic :- Veno-occlusive disease
2. ITP
3. Myeloproliferative diseases
4. Hereditary Spherocytosis
5. SCD (initially)
6. Infectious mononucleosis

CAUSES OF HEPATOSPLEENOMEGALLY:

1. Congenital hepatic fibrosis
2. CF
3. Thalassemia

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4. MPS

COLLECTIONS:

1. Hepatomegaly, no spleen, no scar → SCD
2. Splenectomy scar , no liver, no jaundice , no pallor, +- Kocher incision → Hered. Spherocytosis
3. Splenectomy scar, No liver, Jaundice and pallor +- Kocher incision → SCD
4. Hepatomegaly, periumbilical scar, jaundice , Groin scar → Thalassemia + endoscopic splenectomy
5. Spleen, jaundice , ascitis → Cirrohsis → Portal HTN
6. Huge liver, mild spleen, no jaundice , no pallor → GSD

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Neurology

General obseravation:- as usual + dysarthria ? difficult speech? Deviated mouth? Telangectasia? Squint? Nystagmus? Dysmorphism? Walking aids? Any specific surroundings any specific posture

1. Ask the mother if he can walk
2. Ask the child to walk
3. First :- let him stand for inspection
4. EXPOSE & put off the shoes.
5. Inspection:- muscle bulk, scars? Any obvious deformity, **inspect the shoes**
6. Inspect the back :- any kyphosis, scoliosis, scars
7. GAIT :- عادي، طراطيف صوبعك ، كعب رجليك ، جنب رجليك ، اجري

Spastic gait
Hemiplegia

= Normal speech , may be squint, walking aids,

= **Inspection**:- scars, ~ normal muscle bulk or atrophy

= Spastic gait

= Lye on the bed for motor examination of the LL

= **Tone**:- hyper tonia

= **Power**:- weakness

= **Reflexes**:- hyper reflexia

= **Babeniski**:- dorsi flexion

= **Clonus**:- +ve

= I'd like to examine the motor function of the UL:- Tone , power , reflexes

= In order to complete my examination I'd like to examine the **cranial nerves** and the sensation

Spastic gait
Diplegia

= speech may be affected, may be squint, walking aids, may be affected hearing or vision
= **Inspection**:- scars, ~ normal muscle bulk or atrophy (scar in the groin)

= Spastic gait (Specific gait)

= Lye on the bed for motor examination of the LL

= **Tone**:- hyper tonia

= **Power**:- weakness

= **Reflexes**:- hyper reflexia

= **Babeniski**:- dorsi flexion

= **Clonus**:- +ve

= I'd like to examine the motor function of the UL:- Tone , power , reflexes

= In order to complete my examination I'd like to examine the **cranial nerves** and the

Waddling gait
Duchenne / proximal myopathy

= Normal speech , walking aids,

= **Inspection**:- No scars, hypertrophied calf, relatively small thigh, scar of MS biop.

Back :- may be scoliosis

= Waddling gait, LORDOSIS

= GOWER SIGN +ve

= Lye on the bed for motor examination of the LL

= **Tone**:- hypotonia

= **Power**:- weakness **more proximal**

= **Reflexes**:- absent knee, may be present ankle

= **Babeniski**:- -ve Babeniski sign

= **Clonus**:- don't do

= I'd like to examine the motor function of the UL:- Tone , power , reflexes
proximal weakness

= stick your tongue out :- SMA III

In order to complete:-

- CVS examination → cardiomyopath
- Spirometry→ restrictive lung dis
- Developmental assessment (ment retard)

INVX:- . CPK . Genetic .ECHO

TTT:- . Genetic counselling . ortho . pulm
. Cardiologist . physio . Occup

High steppage gait
peroneal ms atrophy

= Normal speech , may be walking aids,

= **Inspection**:- No scars, inverted chpmaign bottle, distal atrophy, pes cavus

= High steppage gait

= Romberg's +ve

= lye on the bed for Motor examination of the LL

= **Tone**:- hypotonia

= **Power**:- weakness, **more distal**

= **Reflexes**:- hyporeflexia

= **Babeniski**:- -ve

= Clonus:- don't do !

= **Sensation :- Sensory level**

- Lt side of the leg → S1
- Middle 3 toes→ L5
- Big toe → L4
- Lower thigh → L3
- Middle thigh → L2
- Upper thigh → L1
- Symphysis pupis → T12
- In between → T11
- Umbilicus → T10

→ Test position sensation

= I'd like to examine the motor function of the UL:- Tone , power , reflexes

= In order to complete my examination I'd like to examine the cranial nerves and the

Myotonic dystrophy

= **Obs:-** My be problem with speech , walking aids, may be wheel chair bound, Mask face + Ptosis+ Vshaped mouth

= **Inspection**:- No scars, may be muscle atrophy

Back :-

= Gait :-

= GOWER SIGN :-

= Lye on the bed for motor examination of the LL

= **Tone**:- hypotonia

= **Power**:- weakness

= **Reflexes**:- absent knee,

= **Babeniski**:- -ve Babeniski sign

= **Clonus**:- don't do

= I'd like to examine the motor function of the UL:- Tone , power (hand shake the pt or the mother), reflexes

= stick your tongue out :- SMA III

In order to complete:-

- Eye examination:- cataract

INVX:-

- Ggenetic testing
- Screening for DM and cataract

TTT:- . Physio . Occup . Geneticist

Ataxic gait + Abnorm. movements وهوا قاعد

Rh Chorea/ Dyskinetic CP

= Dysarthraia, weak speech, Abnormal movements while seated

= **Inspection**:- No scars, ~ normal muscle bulk or atrophy

= Ataxic gait

= Tandem gait → Stand still,

= Romberg's sign (ask the examiner's permission)

= Motor examination of the LL

= Tone:- +++ = Power:- = Reflexes:-

= **Babeniski**:- = Clonus:- don't do !

= coordination :- heel-knee-chin

= UL examination:-

= Tone:- +++ = Power:- = Reflexes:-

= finger to nose = finger to my finger

= dysdyadokokinesia

→ Abn. Mvmts affecting the coordinatio

= Inoticed some abnormal movements , so I want to do some tests to check for Choreoathetosis:-

- **Milkmade**
- **Spooning**
- **Pronator drift**
- **Jack in the box tongue**

= I'd like to examine the motor function of the UL:- Tone , power , reflexes

= In order to complete my examination I'd like to examine the cranial nerves and the

Unsteady gait / ataxic gait
Ataxia telangectasia/ Freidriech's ataxia
mixed CP/ cerebellar ataxia

= **Difficult speech , may be Dysarthria,** Nystagmus, walking aids,

= **Inspection**:- No scars, ~ normal muscle bulk or atrophy

= Ataxic gait

= Tandem gait → Stand still,

= Romberg's sign (ask the examiner's permission)

= Lye on the bed for motor examination of the LL :-

Ataxia Telangectasia	Freidrich's Ataxia	Mixed CP
Tone:- NAD	**Tone**:- hypotonia	**Tone**:- Hypertonia
Power:- NAD	**Power**:- weakness	**Power**:- weakness
Reflexes:- NAD	**Reflexes**:-Hyporeflexia(ankle)	**Reflexes**:- Hyperreflexia
Babiniski: NAD	**Babiniski**: +ve	**Babiniski**: +ve
Sensation:- NAD	**Sensation:-** Affected	**Sensation:-** NAD
Coordination:- Affected	**Coordination**:- Affected	**Coordination**:- Affected

Cerebellar Ataxia

Tone:- Hypotonia / NAD

Power:- weakness

Reflexes:-Hypo/NAD

Babiniski: -ve

Sensation:- NAD

Coordination:- Affected

= Coordination examination of the UL

= I'd like to examine the motor function of the UL:- Tone , power , reflexes

= In order to complete my examination I'd like to examine the cranial nerves and the sensation

Additional notes

Ataxia Telangectasia / Freidrich's Ataxia/ Ataxic CP

1. Cerebellar ataxia(more common)-ve Romberg's
2. Freidrick's ataxia(hypotonia, hyporeflexia,+BAB.& + ROMBERG'S)>>>> spinocerebellar degeneration(sensory post.colomn)
3. Ataxic CP (Ataxia+Diplegia)

Freidrich's Ataxia

In order to complete my examination:-

- The rest of the neurological examination: e.g:- UL , sensation , cranial .....
- CVS examination → associated with hypertophic cardiomyopathy
- Formal eye examination → associated with optic atrophy

MANAGEMENT:

- **INVX:-**
  - Genetic testing for the gene FXN on chromosome 9
  - ECHO for hypertrophic cardiomyopathy
  - Spine X-Ray for kyphoscoliosis
  - Blood Sugar (associated with DM)

- **MDT:-**
  - Geneticist
  - Paediatric neurologist
  - Cardiologist
  - Orthopaedic surgeon
  - Endocrinologist → associated with DM
  - Physiotherapist
  - Occupational therapist
  - Ophthalmologist → optic atrophy
  - SALT for slurred speech
  - Psychologist
  - Social worker

Prognosis☹FREIDRECK'S:-

- Mainly supportive
- Progressive ataxia and wheelchair bound at mid-twenties, death at 40 or 50 y of age in most cases.
- 75% scoliosis
- Hypertrophic cardiomyopathy, angina, arrhythmia >>> death cause

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• Ataxia telangectasia

In order to complete my examination:-

- The rest of the neurological examination: e.g:- UL , sensation , cranial .....
- Examine the eyes and skin for telangiectasias
- Examine the lymphoreticular system as a screening for malignancy

➢ Management

- **INVX:-**
  - Genetic testing → AR on chromosome 11
  - Immunoglobulins level → low IgA & IgG
  - Alpha-fetoprotein → usually raised
- **MDT:-**
  - Geneticist → for genetic counselling
  - Paediatric neurologist
  - Immunologist for immunoglobulins transfusion
  - Physiotherapist
  - Occupational therapist
  - SALT for slurred speech
  - Psychologist
  - Social worker

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•Hemiplegia

. **Management:**

1. Firstly i would like to take a thorough **history** from the mother in the form of perinatal, natal and postnatal histories, any history of trauma infection or any family history of similar condition, also I want to make a full developmental assessment.
2. **Investigations** in the form of CBC, coagulation profile, protein C&S, and anti thrombin III levels, sickle cell screening and MRA scan.
3. **MDT** in the form of:

.my consultant .neurologist (head of the team) .physiotherap
.Occupational th. .orthopedic surg. .ophthalmologist(+/-)

.ENT (+/-) .speech th.(+/-) .hematologist .dietician .psychologist and school

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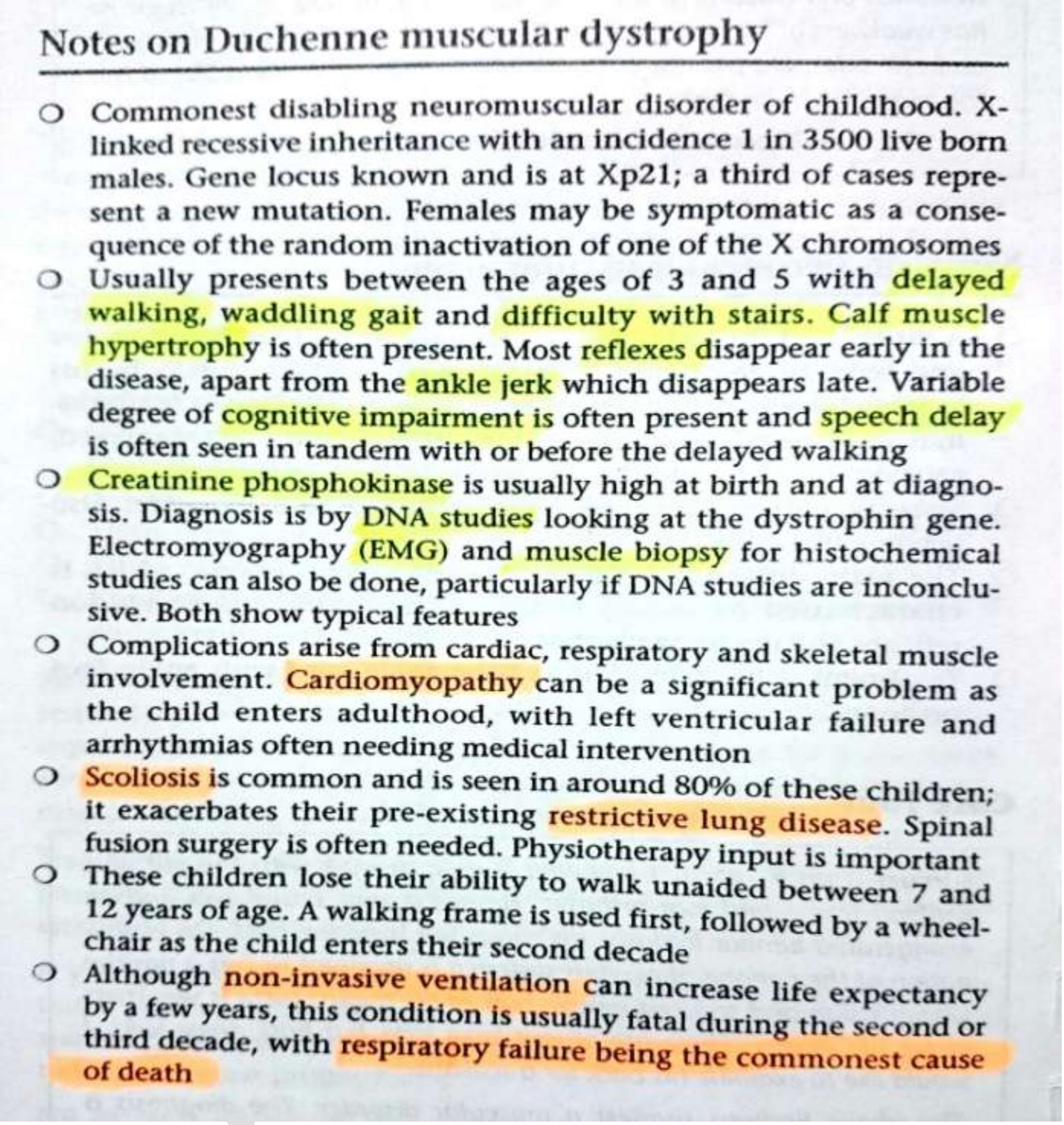
• Duchenne Muscular dystrophy :- →XL



Notes on Duchenne muscular dystrophy

- Commonest disabling neuromuscular disorder of childhood. X-linked recessive inheritance with an incidence 1 in 3500 live born males. Gene locus known and is at Xp21; a third of cases represent a new mutation. Females may be symptomatic as a consequence of the random inactivation of one of the X chromosomes
- Usually presents between the ages of 3 and 5 with delayed walking, waddling gait and difficulty with stairs. Calf muscle hypertrophy is often present. Most reflexes disappear early in the disease, apart from the ankle jerk which disappears late. Variable degree of cognitive impairment is often present and speech delay is often seen in tandem with or before the delayed walking
- Creatinine phosphokinase is usually high at birth and at diagnosis. Diagnosis is by DNA studies looking at the dystrophin gene. Electromyography (EMG) and muscle biopsy for histochemical studies can also be done, particularly if DNA studies are inconclusive. Both show typical features
- Complications arise from cardiac, respiratory and skeletal muscle involvement. Cardiomyopathy can be a significant problem as the child enters adulthood, with left ventricular failure and arrhythmias often needing medical intervention
- Scoliosis is common and is seen in around 80% of these children; it exacerbates their pre-existing restrictive lung disease. Spinal fusion surgery is often needed. Physiotherapy input is important
- These children lose their ability to walk unaided between 7 and 12 years of age. A walking frame is used first, followed by a wheelchair as the child enters their second decade
- Although non-invasive ventilation can increase life expectancy by a few years, this condition is usually fatal during the second or third decade, with respiratory failure being the commonest cause of death

. Management:

1) FIRSTLY I would like to take a thorough history from the mother complete family history of similar condition, making genetic counselling and developmental history.

2) Investigations in the form of genetic test(xp21)>>DNA (decreased dystrophin gene), CPK, EMG,muscle biopsy

3) MDT in the form of:

.my consultant .neurologist (head of the team) .physiotherapy

.Occupational th. .orthopedic surg. .dietician

.psychologist .school .genetist .cardiologist

DD>>> kugelburg wallender(smaiii): → AR

+ Gower's, waddling gait (as DMD)
Hypotonia, hyporeflexia, and decreased PROXIMAL muscle power.
+++++ Winging of the scapula
+++++ tongue fasciculations (lmnl)
DD: with **limb girdle muscle dystrophy** (to differentiate→DNA, EMG and NCS)→ **AR**
DD:- **Facioscapulohumeral muscular dystrophy → AD**

In myotonic dystrophy:

Hold the mother's hand
Expressionless face
Hypotonia , hyporeflexia
Do EMG

Congenital myopathy:

Hypotonic posture
Myopathic expressionless face
Hypotonia , hyporeflexia, weakness
Sensation intact and nothing in the back(# Spina bifida)

Causes of expressionless face:

1. Myopathies
2. Myotonic dystrophy
3. Mitochondrial diseases
4. Myasthenia gravis
5. Fascioscapulohumeral Muscle Dystrophy

HSMN:- ttt → stabilisation of the ankle joint with ankle-foot orthoses

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SPINA BIFIDA

General observation:- Wheel chair, Large head, VP shunt, Hypotonic position, Diaper, catheter, squint, facial deviation.

Lower Limb Examination:-

Inspection:- Wasting, Scars, bed sores, ulcers

Tone:- Hypotonia

Power:- Weakness grade 0-1 ارفع رجلك

- If not able to lift leg (hip flexion) → lesion above L1
- If able to lift leg (hip flexion)→ lesion below L1

Motor leveling:-

- فوق L1--- نايم مش بيقعد
- تحت L1 —ممكن يقعد
- تحت L3 --- يقدر يزحف
- تحت S3--- يقدر يمشي و احساس طبيعي بس incontinent

Reflexes+ Babiniski:- Hyporeflexia / Areflexia

Sensory examination:- sensory leveling

- Peri anal area → S3, S4
- POSTERIORLY>> MED. SURFACE OF THE UPPER CALF→S2
- **Start here** →Lt side of the leg → S1 if not feeling → continue as down to know the level, if feeling → continue as up
- Middle 3 toes→ L5
- Big toe → L4
- Lower thigh → L3
- Middle thigh → L2
- Upper thigh → L1
- Symphysis pupis → T12
- In between → T11
- Umbilicus → T10

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Back:- Scar of spina bifida, Sores, Scoliosis

Head examination:- Head circumference, shunt, sutures

Abdomen:- masses, shunt scar, shunt nephritis, neurogenic bladder

In Order to complete:-

- Abdominal examination (neurogenic bladder, scar of VP shunt)
- Anal examination (patulous anus)
- I need to check the urinary catheter (urethral / suprapupic)
- Check hip for contracture and dislocation

DX:- Neural tube defect causing spina bifida most probably part of Arnold Chiari type II

Advice to the patient:-

- Change position → sores
- Urine analysis → UTI
- Hot plates → burn bcz no sensation
- if he is going to surgical operation I should advise him that he may have LATEX ALLERGY

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- **Discussion:**
  I examined a 6 y old boy he is looking small for his age so I want to plot his measurements on the appropriate growth chart.
  Ahmed has a hypotonic posture, unable to sit or move the lower part of his body in the form of hypotonia and hyporeflexia with decrease muscle power and negative Babiniski sign, he has a scar in his lower back with shunt from his head reaching the Rt hypochondruim. So I believe this is a case of operated spina bifida most commonly due to Arnold Chiari type II

. MANAGEMENT:

1)FIRSTLY I would like to take a thorough **history** from the mother(folic) complete family history .

2)**Investigations** in the form of CT, MRI and urine dipstick to exclude UTI

3)**MDT** in the form of: .my consultant .neurologist (head of the team) .physiotherapy .Occupational th. .orthopedic surg. .community ped. .dietician .psychologist .school .Urologist (Intermittent cath.) and antibiotic prophylaxis

COMPLICATION OF SHUNT:

- Infection
- Obstruction
- Slit ventricle syndrome(over drainage>>headache)
- Peritonitis or fibrosis

SENSATION

- Inguinal region>>l1
- Med. Surface of the upper thigh>>l2
- Med. Surface of the lower thigh>>l3
- Med. Surface of the leg>>>>l4
- Lat. Surface of the leg>>>>l5
- Lat. Surface of the foot>>>s1
- Then >>> posteriorly>> med. Surface of the upper calf>>.s2
- Med. Surface of the inner thigh s3
- Perianal area>>>s4

REFLEXES:- **BICEPS:-** C5,C6 **TRICEPS:-** C7,C8 **KNEE:-** L3,L4 **ANKLE:-** S1,S2

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SMA III

Like Spina Bifida BUT:-

- Normal IQ
- Small for age
- Not moving the UL
- Tongue fasciculation in 60%
- No back scar
- Intact sensation
- In order → chest examination , spirometry
- Diagnosis → gene study , EMG

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Werding Hoffman

Infant Approach

General observations:- as usual + posture (frog leg position, paucity of movement , awareness, following? Infant, Alert, moving his eyes interested in the surroundings, frog leg position, Paucity of movements, bell shaped chest, paradoxical sea saw respiration

EXAMINATION:-

- open mouth to see the fasciculations.
- Head Support
- Truncal Support
- Vertical Suspension
- Ventral Suspension
- Prone position
- Sitting ?
- Standing ?
- TONE, POWER, REFLEXES (don't do babiniski !!)

- Finally examine the persistence of the primitive reflexes
  - Moro Reflex (ask parent's permission)
  - Tonic Neck Reflex
  - Grasp Reflex
  - Parachute Reflex

- **DD:** SMA, HYPOTONIA FOR DD, CP.

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QUADRIPLEGIC CEREBRAL PALSY

- **General observation:-**

  - Talk to the child (raport and dysarthria), YES TALK THE CHILD (many of these patients are intelligent, also exclude hearing loss, speech disorder, visual loss)
  - Well, I'm examining a ……, he is not able to follow me so I would like to make a formal visual assessment at the end of my examination; also I would like to assess his hearing and speech. He has a windswept deformity, multiple contractures; he has an apparently small head I would like also to put his HC on the appropriate Growth chart.
  - Comment on teeth, drooling or anything abnormal in his face(adenoid facies or abnormal respiratory noises or snoring if he is sleeping), shunt…
  - wheel chair???? or …….

- **Examination:-**
  - **Inspection:-**

    1. Symmetry in length (difficult due to deformities, windswept….., inversion….adduction)
    2. Muscle bulk (decreased)
    3. Scars??..... It will be difficult to assess.
    4. Pigmentation
    5. gastrostomy

  - **Back examination:-** (scoliosis), nappies, , bed sores…..
- **LL Motor Examination :**
  1. Tone (increased)
  2. Muscle power(decreased)
  3. Reflexes(exaggerated, but could be lost in severe deformities),

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4. Clonus and babiniski exaggerated.

- **Upper limb examination:**
  Same as LL hypertonia, hyperreflexia with decrease muscle power

- **Thank the child or the mother and cover him**

- **At the end of examination I would like to examine for hearing, vision and speech and cranial nerves**
- **N.B. if associated With choreoathetoid mov.>> lesion involved the extrapyramidal tract.(Kernectirus).**

➢ Discussion:-

I examined a 6 y old boy he is looking small for his age so I want to plot his measurements on the appropriate growth chart.

Ahmed has a windswept deformity(risk for hip dislocation) with multiple flexion deformities in LL and UL, hypertonia, hyperreflexia, Bab. +,with apparently small head, he was not responding to me , and was not able to follow my hands, so I have to check his vision, hearing and speech. So this is a case of UMNL in the form of **quadriplegic CP.**

➢ MANAGEMENT:

1) FIRSTLY I would like to take a thorough history from the mother as pre natal, natal and postnatal.

2) Investigations: CT, MRI, vision , hearing and full developemental assessment.

3) MDT in the form of:

.my consultant
.Occupational th.
.dietician
. Support groups
.ENT
.neurologist (head of the team)
.orthopedic surg.
.psychologist
. Social worker
.Respite care
.physiotherapy
.community ped.
. Sp.school
.opthalmologist

Types of CP: (a) SPASTIC (b)ATAXIC (c) DYSKINETIC

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CRANIAL NERVES

I.OLFACTORY :- Ask about sense of smell

II. OPTIC:- (Inspect the eye first)

- Visual acuity:- Establish if the child can see (each eye separately):- count finger من بعيد و بعدين قريب, hand movement, light perception
- Accommodation
- Visual field:- confrontation test (الاول و العينتين الاتنين مفتوحين و بعد كده عين عين)

III, IV, VI occulomotor, Trochlear, Abducent:-

- أحط ايدي علي أورته علشان أثبت راسه، اقوله خليك باصص علي صباعي و تابعه من غير ما تحرك راسك ولو شوفت القلم اتنين قوللي، احرك القلم في كل الاتجاهات (H shaped) وبعدين أعمل العين التانية.
- Check for nystagmus, normally present at extremes of gaze

V. TRIGEMINAL:-

- SENSORY:-
  - غمض عينيك
  - لما تحس اني بلمسك قول نعم
  - ألمسه بقطنة في أورته ، خده ودقنه في النحيتين
- MOTOR
  - افتح بقك وشوف لو فيه deviation
  - اقفل بقك وجز علي اسنانك
  - احس بأيدي ال TEMPORALIS & MASSETER

VII. FACIAL:-

- ارفع حواجبك
- اقفل عينيك جامد
- ماتخلينيش افتحهم
- وريني اسنانك
- صفر
- انفخ بقك

VIII. VESTIBULO-AURICULAR:-

- اقفل ودن واهمس في الودن التانية رقم واسأله عليه

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IX. GLOSSOPHARYNGEAL:-

- Gag reflex قوله ماتعملوش (رايح في 9 وراجع في 10)

X. VAGUS:-

- يفتح بقه ويقول آه :- Deviated uvula if paralysed
- يتكلم :- hoars voice if paralysed

XI. ACCESSORY:-

- أحط ايدي على دقنه وأقوله زق ايدي
- أحس بالإيد التانية ال Sternomastoid في عكس اتجاه الزق
- ارفع كتافك ومتخلنيش انزلهم

XII. HYPOGLOSSAL:-

- طلّع لسانك
- حرّكه يمين وشمال زق علي ايدي شمال و يمين

In order to complete:-

- Test :- smell , taste, Gag reflex, light reflex, jaw reflex and corneal reflex
- do rinne and weber test
- Examine the patient's glasses
- Fundus examination
- Full neurological examination

FACIAL NERVE PALSY

- ارفع حواجبك
- اقفل عينيك جامد
- ماتخلينيش افتحهم
- وريني اسنانك
- صفر
- انفخ

In order to complete:-

- Examine Taste sensation
- BP

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- Ear examination
- Inspect skin for rash (erythema migrans)
- Assess hearing
- Examine other cranial nerves
- Full neurological examination of the upper and lower limbs

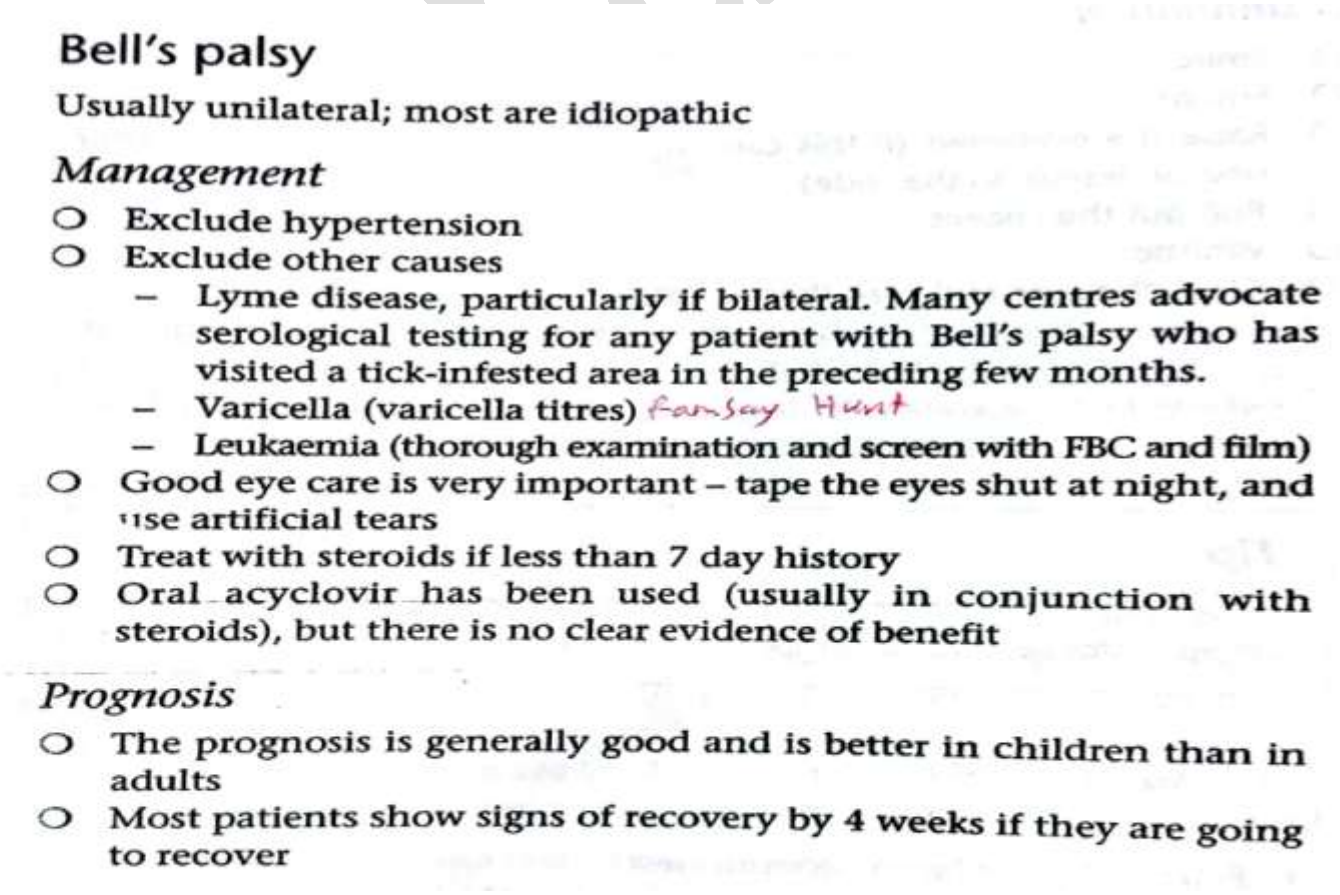
AETIOLOGY OF FACIAL NERVE PALSY:

(a) LMNL:

. Bell's (the commonest)
. Ramsy hunt syndrome(HZV)
.Chronic serious OM
.intracranial tumors
. Viral infections, Lyme
. Congenital
.GB syndrome
. Trauma

(a) UMNL

. CP
. Tumors - Mobuis syndrome (strabismus and immobile face)



Bell's palsy

Usually unilateral; most are idiopathic

Management

○ Exclude hypertension
○ Exclude other causes
  – Lyme disease, particularly if bilateral. Many centres advocate serological testing for any patient with Bell's palsy who has visited a tick-infested area in the preceding few months.
  – Varicella (varicella titres) Ramsay Hunt
  – Leukaemia (thorough examination and screen with FBC and film)
○ Good eye care is very important – tape the eyes shut at night, and use artificial tears
○ Treat with steroids if less than 7 day history
○ Oral acyclovir has been used (usually in conjunction with steroids), but there is no clear evidence of benefit

Prognosis

○ The prognosis is generally good and is better in children than in adults
○ Most patients show signs of recovery by 4 weeks if they are going to recover

EYE EXAMINATION

General observation:- Obvious squint, ptosis, Nystagmus, dysmorphism, glasses, Any head tilt

Look at the eyes:-

- Conjunctiva
- Pupils (any corneal opacities)
- Lids
- Check for nystagmus:- normally present in the extremes of gaze

visual acuity:-

- Check if the child can see :- Reading, count finger, hand movement, light perception.
- Test both eyes separately
- By the end of the exam I’d like to do a formal visual assessment.

Accommodation:-

- أنا هاقرب صباعي منك، عاوزك تفضل باصص عليه
- Observe for convergent movement of the eyes and pupils constriction

Visual field test:- confrontation test

Eye movement:-

III, IV, VI occulomotor, Trochlear, Abducent:-

- أحط ايدي علي أورته علشان أثبت راسه، اقوله خليك باصص علي صباعي و تابعه من غير ما تحرك راسك ولو شوفت القلم اتنين قوللي، احرك القلم في كل الاتجاهات (H shaped) وبعدين أعمل العين التانية.

Check for nystagmus, normally present at extremes of gaze

Light reflex:-

- احط ال torch قدام عينه و أشوف ال pupil constriction في نفس العين (direct) و في العين التانية (consensual)

Squint examination:- → Page 310 Mark Beattie

- Corneal reflection
- Eye movement → if not done

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- Cover/Uncover test
- Alternate cover test

In order to complete:-

- Examine the patient's glasses
- Fundus examination
- Full neurological examination including cranial nerves

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Non paralytic>> usually due to refractory error
Paralytic >> usually due to muscle problem.

Management:-

- Ophthalmologist
- Optometrist
- Orthoptist
- Brain imaging

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visual assessment by age:-

= 3 years → optokinetic Nystagmus(circular moving tissue)
= 3-4 years → matching V-Chart(letters)
= >5 years → snellen’s chart

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Erb’s Palsy

Affected muscles:-

- Deltoid
- Biceps
- Brachioradialis
- Brachialis

Presentation:-

- Shoulder → Adduction + internal rotation
- Elbow → Extension + pronation
- Wrist → Flextion
- Fingers → maintained grasp

Opening statement :- difficult writing , difficulty using hands , …..

Observation:- as usual + specific position

General Examination:- Inspection :- for wasting, deformities, scar , look under the axilla for scar of repair

Local examination:-

- **Tone**:- hypotonia
- **Power**:- weakness :- مش قادر يعمل وضع البطلة ولا ال Boxing بالمقارنة بالايد التانية
- **Reflexes** :- Hyporeflexia
- **Sensation loss over**:-
  - Thumb → C6
  - Tip of the index finger
  - Outer half of the Arm and Forearm

In order to complete my examination:-

- Eye examination because sometime associated with Horner’s syndrome
- Motor examination of the LL and cranial nerves

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Additional notes

Difference between 3rd nerve palsy and Horner:-

= Horner → Ptosis + Miosis + No squint

= 3rd nerve palsy → ptosis+ Mydryasis + Squint (outward downward deviation of the eye) + -ve light reflex and accommodation

Ulnar nerve affection

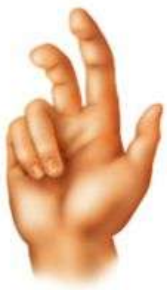
- Claw hand
- Loss of sensation over the medial 1 ½ fingers of the hand on the palmer and dorsal sides
- Spared thenar muscles
- Not able to extend muscles at the level of interphalangeal joints

Radial nerve affection

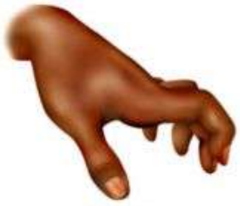
- Wrist drop
- Sensation lost at the anatomical snuff box
- Absent like sign

Median nerve affection

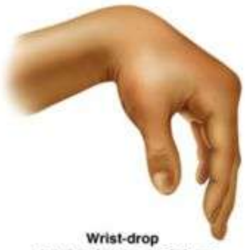
- Ape's hand
- Wasting of thenar muscles
- Loss of sensation at the lateral 3 ½ fingers on the palmer side
- Able to flex the medial 2 fingers



"Hand of benediction" Or ape hand
(a) Median nerve injury



Clawhand
(b) Ulnar nerve injury



Wrist-drop
(c) Radial nerve injury

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MSK UPPER Limb

General Observ

- obvious deformity, posture , well grown?
- obvious dysmorphism
- Aids:- wheel chair, orthoses, crutches

Screening Quest

- عندك وجع في عضلاتك او مفاصلك؟
- فيه اي وجع في ايدك؟
- فيه مشكلة ف طلوعك ونزولك السلم

LOOK

- **D**eformity
  - Arachnodactyly (marfan)
  - Short 4th & 5th metacarpals (turner)
- **A**symmetry
- **M**uscles
  - Wasting in thenar or hypothenar
- **P**osture
  - Claw hands(brachial plexus inj medial cord)
  - Flexed hand and arm (hemiplegia)
  - Wrist drop (radial palsy)
  - Ulnar deviation (rheumatoid arthritis)
- **N**ails
  - Clubbing
  - Pitting, onycholysis (psoriasis)
  - Koilonychias (iron deficiency anemia)
- **S**kin
  - Eczema
  - Psoriatic rash
  - Gottrons papules (dermatomyositis)
  - Any other rash
- **S**tigmata
  - Neurocutaneous stigmata
  - Stigmata of bacterial endocarditis
- **S**cars

Feel

- Ask about pain in the hands
- Temperature and sweating
- Palpate any swelling to know if it's bony or soft
- Squeeze metacarpal joints for tenderness

Move

Check for Hypermobility

- افرد صوابعك make a star
- Make a fist
- اقلب ايدك supination
- Make a circle (opposition)
- عد صوابعك
- Palms then backs
- If there is any limitation of movement, say that you’d like to measure it formally using goniometer

Measure

- افرد ايديك الاتنين قصاد بعض
- Do mid arm circumference measurements

Joint above and joint below

Elbow in case of hand affection

- Look, Feel, Move , measurements

For the Elbow :-

- Move →
    - Flexion
    - Extension
    - Pronation
    - Supination

For the Shoulder:-

- Move →
    - ارفع ايدك للسما
    - حط ايدك ورا راسك
    - حط ايدك ورا ضهرك

Function

- اكتب
- اشرب
- سرح شعرك

In order to complete

- PGALS screening of the other joins
- Associations (extra articular manifestations):-
    - Eyes:- blue sclera, visual impairment from uveitis, conjunctival pallor (anaemia)
    - Skin:- psoriatic lesions, dermatomyositis rash, stria from steroid use, neurocutaneous stigmata, SLE rash, Salmon pink rash
    - Nails:- Psoriatic changes, iron deficiency changes
    - Abdomen:- Hepatosplenomegaly
    - Look for overgrowth or undergrowth of the affected limb (eg:- measure for length discrepancy)
    - Check child's temperature (systemic onset JIA)

- Complications of medical therapy
- If there is scoliosis:-
    - look for cutaneous stigmata
    - Respiratory examination+ Respiratory function tests
    - Neurological examination

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JIA

INVX:-

- Immune labs:- RF, ANA, anti Dst DNA,
- Search for other autoimmune associations:- RBS, celiac screen
- X-ray

Management:-

- NSAIDs :- Ibuprofen + Antacids
- Steroid injection , systemic steroids
- disease modifying medications (methotrexate), Anti-TNF monoclonal AB (infliximab)

- physiotherapist and Occupational therapist for :- hydrotherapy, passive movements, gentle exercises and splints.

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MSK LOWER Limb

General Observ	• obvious deformity • obvious dysmorphism • Aids:- wheel chair, orthoses, crutches, Special shoes
Screening Quest	• عندك وجع في عضلاتك او مفاصلك؟ • فيه اي مشكلة في طلوعك ونزولك السلم؟ • فيه اي مشكلة في انك تقلع او تلبس هدومك بنفسك؟
Expose lower limbs	
Stand → (esp from back of the LL)**look for**	• Deformity (genu valgum, genu varus) • Joint swelling • Muscle wasting • Leg length inequality • Flat feet + Hyper extended knees • Rash (for psoriasis) • Scar • Back (kyphosis) بص من الجنب
ميل لقدام والمس الأرض بايديك	• Look at the back for scoliosis + spine hyper laxity
Walk, Heel, tip toe, side	• For gait abnormality, Squat, stand on one leg alternatively
Lye on the bed	• If there is no obvious Arthritis, do PGALS, ,, if there is, then do local Joint examination (Look, feel, move)
Look	• Deformity (genu valgum, genu varus) • Joint swelling + measure if it's there • Muscle wasting • Leg length inequality • Rash (for psoriasis) • Nail Pitting
Feel	• Tenderness • Raised local temperature • Effusion (patellar tab + cross fluctuation)

Move

- **Active movement → look for reduced range of movement + feel for crepitus**
- **Passive movement → for painful movement**
- **Put your hand under the knee to check if there is any limitation of extension**
- **If there is any limitation of movement, say that you'd like to measure it formally using goniometer**

Measure

- **Leg length**
- **Quadriceps diameter**

Joint above and joint below

- **Look, Feel, Move , measurements**

For the Hip :-

- Look → skip because it's covered
- Feel → do pressure only over the clothes for tenderness
- Move →
    - Flexion
    - Internal rotation
    - External rotation
    - Abduction
    - Adduction

For the Ankle:-

- Move → planter flexion, Dorsiflexion, inversion, eversion

Associations

- **Heamophilia → cannulation sites, bruises, permacath**
- **JIA, RA :- eye, Abdomen, Rash**

Ab

In order to complete

- PGALS screening of the other joins
- Associations (extra articular manifestations):-
  - Eyes:- blue sclera, visual impairment from uveitis, conjunctival pallor (anaemia)
  - Skin:- psoriatic lesions, dermatomyositis rash, stria from steroid use, neurocutaneous stigmata, SLE rash, Salmon pink rash
  - Nails:- Psoriatic changes, iron deficiency changes
  - Abdomen:- Hepatosplenomegaly
  - Check child's temperature (systemic onset JIA)

- Complications of medical therapy
- If there is scoliosis:-
  - look for cutaneous stigmata
  - Respiratory examination+ Respiratory function tests
  - Neurological examination
  - Evidence of Marfan's syndrome if the child is tall

Example of a case of arthritis

I examined Yasmine , she has an abnormal gait, no scoliosis, there is a swollen left knee with flexion position. There limitation of joint movement with limited extension compared to the rt knee. Also I believe that there is joint effusion. My impression is that she has a chronic inflammatory arthritis (DD infection, infiltration, trauma, tumor, bleeding>> **hemophilia** in boys, JIA, STILL'S, SLE, HSP…>> COLLAGEN DISEASES)

1. I want to review his previous notes
2. **Investigations**: CBC, inflammatory markers, CRP, ESR, ANA, other autoimmune Abs , coagulation profile, factor 8 & 9 assays and imaging in the form of x-ray, sonar and MRI.
+ Genetic testing for heamophilia.
3. MDT:
- ✓ My consultant
- ✓ RHEUMATOLOGIST
- ✓ PHYSIOTHERAPY
- ✓ Heamatologist

- ✓ OCC. THEURAPY
- ✓ Orthopedics
- ✓ GP
- ✓ Psychologist
- ✓ School

N.B. medications used in JIA >>NSAID, STERIODS, METHOTREXATE, CYCLOSPORIN, SULFASALAZINE

Advice for hemophilia:-

- ✓ Avoid trauma+ avoid vigorous exercises
- ✓ Hepatitis B vaccine

How to manage factor 8 resistance:-

- ✓ Immunomodulators + Factor 7 transfusion

If factor 8 is not available :- plasma + Cryo transfusion

Factor 8 dose regimen:-

- ✓ Twice weekly in summer, thrice weekly in winter

CAUSES OF SCOLIOSIS:

1. IDIOPATHIC: increased during pregnancy
2. Neuromuscular weakness e.g. DMD
3. Spine or bone problems: Klipel feil, rickets, OI.
4. Syndromes: NF(café au lait spots), MPS.

ABNORMAL RADIUS

- TAR
- FANCONI(microcephaly, microphthalmia, café au lait, renal problems, blood problems)
- HOLT-ORAM
- VACTERL

Abraam H

RICKETS(as MSK)

Can come as general examination (short stature) or MSK examination

General observation:- as usual + large head, obvious deformity + short stature

Screening Questions :-

Expose LL

Stand (legs and back):- look (deformities , genu varum, pes plannus , hyperextended knees, kyphosis, scoliosis)

Bend forward :- for the scoliosis and for hypermobility

Walk :- for any abnormal gait

Lye on the bed :- for PGALs because there was no obvious joint affection

PGALs of the LL:-

1. **Hip**
    1. Flexion
    2. Internal rotation →passive لو معرفش ما تضيعش وقت و اعمله
    3. External rotation → passive لو معرفش ما تضيعش وقت و اعمله
2. **Knee**
    1. Flexion → ده آخرك ؟ وشوف ال angle
    2. Extension → جط ايدك تحت ركبته
    3. Patellar tap + cross fluctuation
3. **Ankle**
    1. Dorsiflexion
    2. Planter flexion
    3. Effusion → بالمرة شوف ال Marfan sign

→ after doing PGALs I couldn’t appreciate joint affection , however I can see some signs of rickets like ……….. …. So I would like to continue my general examination to check for other signs of Rickets.

Abraam H

Hands:-

- Broadening of wrist and elbow joints
- Hypermobility of the Thumb, little finger and elbow.
- Obvious limb deformities from previous fractures.
- Tanner staging at the axilla if at puberty

Head & Neck:-

- Shape
    - Frontal bossing
    - Box shaped head
    - Alopecia totalis → in Vitamin D dependent rickets type 2
- Size → Measure the head circumference (فك الضفيرة !!)
- Sutures → any prominent sutures
- Scars → any scars
- Mouth → teeth

Chest:-

- Rosary beads (ارفع ايدك فوق راسك وميل لناحية)
- Hrrison's sulci
- pigeon shaped chest (pectus carinatum)

Abdomen:-

- examine the liver → Hepatic rickets

Differential diagnosis:-

1. The obvious reason after examination :-
    a. Hypophosphatemic rickets
    b. Vitamin D dependent rickets type 2 → Alopecia totalis
2. Hepatic rickets
3. Renal rickets
4. The other Types:-

a. Vitamin D-related rickets

i. Vitamin D deficiency

Abraam H

        1. Nutritional→(the commonest cause)→ poor sun exposure, poor feeding.
        2. GIT loss:- celiac, cystic fibrosis, steatorrhea (poor absorbtion)
    ii. **Vitamin D-dependent rickets**
        1. Type 1 (vit D 1-alpha-hydroxylase deficiency)→ active vit D deficiency
        2. Type 2 (calcitriol receptor mutation)→ receptor problem

b. **Hypocalcemia-related rickets**
    i. Hypocalcemia
    ii. **Chronic renal failure** → renalosteodystrophy (dec Ca, incr PTH → Bone degradation

c. **Hypophosphatemia-related rickets**
    i. **Congenital**
        1. Vitamin D-resistant rickets (X- linked hypophosphatemic rickets) → decreased Ph reabsorbtion + decreased vit D synthesis, so TTT → oral Ph + Calcitriol
        2. Autosomal dominant hypophosphatemic rickets(ADHR)
        3. Autosomal recessive hypophosphatemic rickets (ARHR)
    ii. **Secondary Hypophosphatemia** (typically secondary to malabsorption)
    iii. **Fanconi's syndrome** → ph loss from the proximal tubules

MANAGEMENT:-

1. I would like firstly to take a thorough history especially nutritional history & family H.
2. Investigations will include :-
    a. bone profile in the form of Ca, Ph, ALP, PTH, Vitamin D level
    b. X-ray of lower ends of long bones>> widening epiphyseal plates, cupping and fraying together with increased joint space.
    c. Genetic testing for inherited reasons
3. Finally I would like to involve a **MDT**:

- Occupational therapist, My consultant, Orthopedic(in severe cases), Health visitor, Radiologist(green sticks are common), Endocrinologist, Geneticist for counselling

Osteogenesis Imperfecta

General observation:- as usual + May be Bed ridden (ask for wheel chair)+ obvious deformities+ obvious wasting , May be sitting with mild deformities

Screening Questions :-

Expose LL

If Moving :- (rarely) → **PGALs screening like up**

If Not Moving :-

Look :- For the deformity , wasting , leg length inequality, any scars back of the joints

PGALs of the LL :- start by movement :- ask him to do active movement → then tell the examiner :- he couldn't do the active movement of PGALs , would you like me to do passive movements?

→ i have seen Ahmad 10 years old child , and on examination there was …….. …………… all these findings keep with the diagnosis of Osteogenesis Imperfecta so I'd like to continue my general examination to check for other signs of OI

Examination:-

Upper limb:-

- Deformities
- Thin skin
- Short limbs
- Bruises
- Scars

Head and Neck:-

- Blue sclera
- Soft scull
- Sutures and fontanels
- Triangular face
- Soft discoloured brittle teeth

- At the end of my examination i want to examine hearing bcz high possibility of hearing loss due to otosclerosis

Chest:-

- Barrel shaped chest

Lower limbs:-

- Deformities
- Thin skin
- Short limbs
- Bruises
- Scars

Back:- offer to examine the back for deformities

tell your positive findings and conclude>> I believe this is a case of skeletal dysplasia mostly a case of OI.

MANAGEMENT:-

4. I would like firstly to take a thorough history especially family H.
5. Investigations will include :-
    a. Dexa Scan
    b. x-ray→ generalized osteopenia + healed old fractures + wormian bones in the scull
    c. Genetic testing
6. Finally I would like to involve a **MDT**:

- Occupational therapist, My consultant, Orthopedic surgeon for aggressive ttt of fractures to avoid future deformities, Geneticist for counseling , social services, Audiologist for Hearing assessment.

7. Medical ttt :- Bisphosphonate which is osteoclast inhibitor

Types:- 2&3 → Severe and recessive ,, 1&4 → Mild and Dominant

Advice :-

- Minimal handling
- Aggressive orthopeadic ttt of fractures to avoid deformities
- Fu hearing and vision

braam H

Arthrogryposis

General observation:- as usual + fixed flexion deformities of the limbs, may be muscle wasting

Screening Questions :-

Expose LL

Stand (legs and back):- look (flexion deformities, scoliosis)

Bend forward :- for the scoliosis

Walk :- for any abnormal gait (may not be able to walk on heels)

Lye on the bed :- for PGALs

PGALs of the LL:-

4. **Hip**
    4. Flexion
    5. Internal rotation →passive لو معرفش ما تضيعش وقت و اعمله
    6. External rotation → passive لو معرفش ما تضيعش وقت و اعمله
5. **Knee**
    4. Flection → angle ده آخرك ؟ وشوف ال
    5. Extension → جط ايدك تحت ركبته
    6. Patellar tap + cross fluctuation
6. **Ankle**
    4. Dorsiflexion
    5. Planter flexion
    6. Effusion → Marfan sign بالمرة شوف ال

PGALs of the UL:-

- Stretch out your hands
- Make a star
- Make a fist
- Supinate
- Make a circle
- Count your fingers
- Palms together

Abraam H

- Backs together
- ايديك للسما
- ايديك ورا راسك
- ايديك ورا ضهرك

In order to complete :- PGALs of the back

Management :-

1. Revise the pt's notes
2. **INVX:-** CK, EMG, Muscle biopsy, Genetic testing
3. **MDT:-** Physiotherapist , Occupational therapist, orthopedic surgeon for splinting and surgery

Arthrogryposis :- congenital joint contracture in two or more areas of the body.

Types:-

- **Amyoplasia** → characterized by severe joint contractures and muscle weakness
- **Distal Arthrogryposis** → mainly involves the hands and feet
- **Syndromic** → Types of arthrogryposis with a primary neurological or muscle disease belong to the syndromic group.

Prognosis:- if not syndromic, can be treated by surgery and so lead a normal quality of life

Examine the Back

Look

- Deformity / scolisis

Feel

- Temperature
- Tenderness
- Test for scoliosis (to differentiate between Positional , Apparent and Fixed)
  - bend forward
  - sit on chair

Move → 6 Back + 6 Neck

- ميل قدام و ورا
- ميل للجنب يمين و شمال

- Twist right and left
- حركات الرقبة

Measure:-

- Angle by Genometry → if >45 degrees → needs surgery
- Spirometry

Dermatomyositis

Skin cases :- SLE, Psoriasis, Dermatomyositis

They may have associated **Arthritis** → Do Local Joint examination

If **no Arthritis** → Do PGALs → I noticed some signs suggesting that the case is Dermatomyosistis so I need to do general examination to look for other signs of dermatomyositis

Hands:- Gottron Pappules , rash on the Arm or Forearm

Head:- Heliotrope rash

Trunk:- Rash

In order to complete my examination :- Neurological examination to check for proximal muscle weakness

Management :-

- **INVX:-**
    - CK , Autoantibodies e.g. ANA
    - EMG
    - Muscle biobsy
- **MDT :-**
    - Physiotherapist
    - Occupational therapist
    - Rheumatologist
- Steroids, Azathioprine , Methotrexate

Advice:-

- Avoid sun exposure
- Avoid sleeping in supine position , Because palatal dysfunction→risk of aspiration and suffocation

THYROID

General observation:- As usual + obvious eye signs, obvious swelling ,any hearing aids (pendred syndrome), any voice hoarseness (especially if there is a scar)

Examination:- Do Justified Exposure

Inspection :- (front with neck extension)

- Neck swelling: - Site, Shape (uniform, unilateral?) , Size, Scar? ,
- Swallowing: - does it move with deglutition?
- Stick your tongue out: - (if it rises during the manoeuvre → it's a thyroglossal cyst)
- Look at the base of the tongue for high thyroglossal cyst

Palpation:- (from back, reassure the pt, ask her to tell if there is pain)push lobe and feel the other

- Neck swelling:- Shape (uniform, unilateral?) , Surface, consistency ,
- Swallowing: - does it move with deglutition?
- Palpate for neck lymph nodes
- Trachea
- Thrill
- Sternomastoid

Percussion:-

- Percuss the sternum for retro-sternal extension

Auscultation:-

- Auscultate the mass for bruit at the edges of the lobes

Then ask the examiner to assess the thyroid status

Hands:-

- Hold hands out , look for tremors
- Look for clubbing
- Feel for raised temperature, sweaty hands
- Pulse :- rate and rhythm and volume, water hammer pulse

Abraam H

- Power حركة البطلة
- I'd like to measure BP at the end of my examination

EYES:-

- Eye brow → loss of outer third of hair
- Lid oedema
- Lid retraction (rim of sclera is seen above the iris) من الجنب
- Exophthalmos (examine from above) & (comment if asymmetrical)
- Lid lag
- Eye movement for External ophthalmoplegia

Dentition:-

- Delayed in Hypothyroidism

Lower limbs:-

- Reflexes(Ankle jerk):- slow relaxation in hypothyroidism
- Proximal myopathy in hyperthyroidism :- (can't get up from squatting position)
- Pretibial myxedema:- hard non pitting edema

Questions to ask to help assess thyroid status:-

- بتحب البرد ولا الحر؟
- وزنك بيزيد و لا بيقل؟
- فيه اسهال ولا امساك؟
- Muscle weakness
- Energy levels
- Schoolwork (usually well behaved if hypothyroid)

In order to complete my examination:-

- Tanner staging
- BP
- CVS
- Abdominal examination for hernia or constipation
- Skin examination for Vitilligo as association

Comment on positive signs and comment whether the pt is clinically hyper/hypo /euthyroid

Put In Your Mind Turner And Autoimmune In A Short Hypothyroid Girl

Abraam H

Why thyroid?

= in the Ant triangle of the neck medial to the sternomastoid
= Anatomical shape of the thyroid, butterfly and isthmus
= move with deglutition
= Doesn’t move with movement of the tongue

MANAGEMENT:-

1. Thorough history
2. Investigations>> TSH, T4, Thyroid stimulating Antibodies, Antithyroid peroxidase, screening for associated autoimmune diseases (celiac, DM→ RBS)
3. **MDT:.** My consultant .Endocrinologist . Opthalmologist . Dietitian .Psychologist .School . Immunologist (autoimmune) . Genetic (Turner)

Treatment of hyperthyroidism:

1. Medical>>carbimazole, propranolol or block therapy>> carbimazole and thyroxine
2. Radioiodine
3. Surgery
4. Prednisolone for eye signs, N.B.>> eye signs usually persist

Treatment of hypothyroidism: L-Troxin

Complications of exopthalmus:

1. Exposure keratitis
2. Optic atrophy(due to compression)
3. Opthalmoplegia (due to lymph. Infiltration).

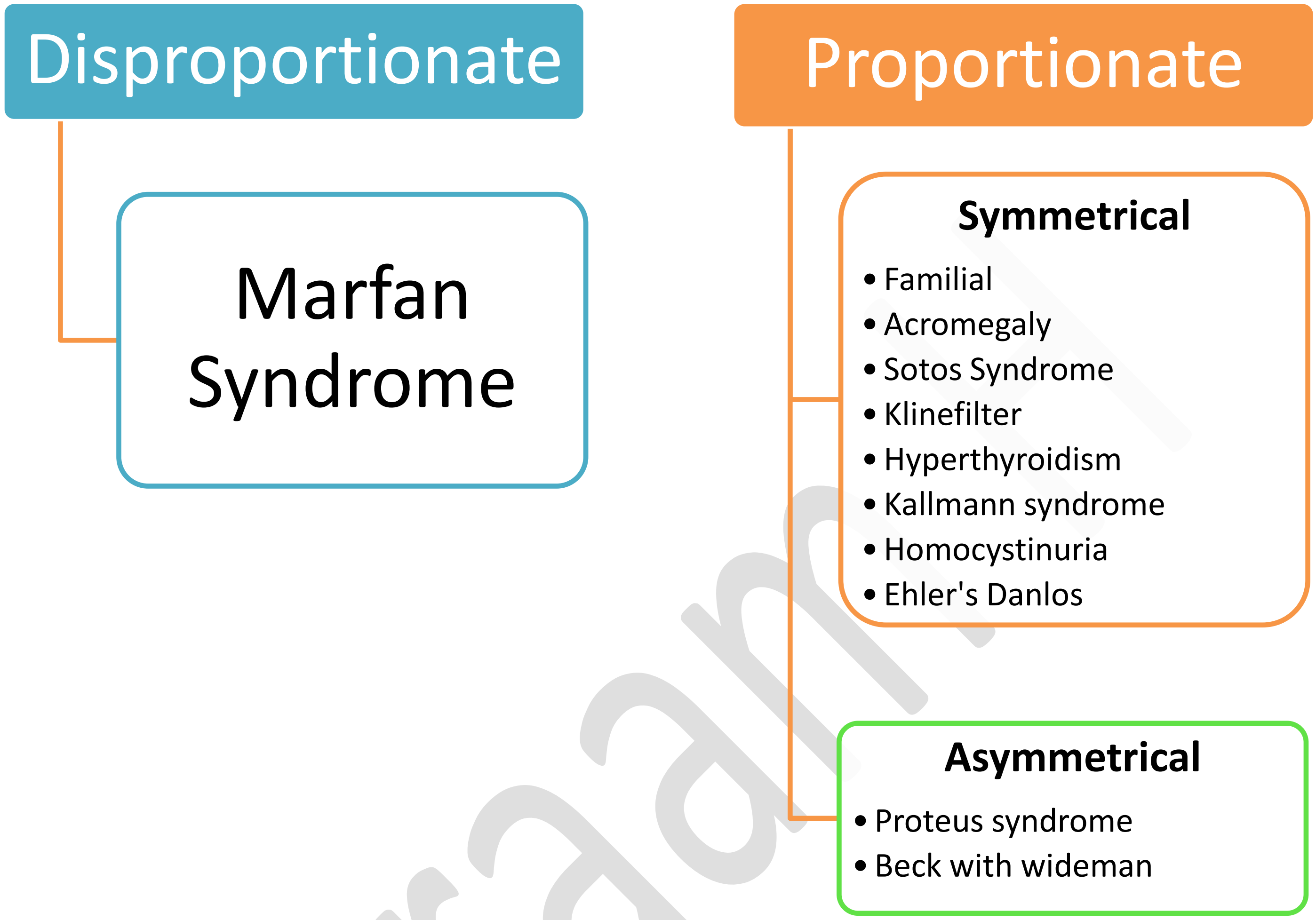
. Hypothyroidism may be associated with Turner, autoimmune disease or Down syndrome.

Head examination:-

- Size
- Shape
- Scars
- Sutures
- Hair
- 3 organs :- eyes , nose , ears
- Cranial nerves

Abraam H

TALL STATURE



Disproportionate

Marfan Syndrome

Proportionate

Symmetrical

- Familial
- Acromegaly
- Sotos Syndrome
- Klinefilter
- Hyperthyroidism
- Kallmann syndrome
- Homocystinuria
- Ehler's Danlos

Asymmetrical

- Proteus syndrome
- Beck with wideman

GENERAL:- as usual + looks Tall , I would like to take his measurements :-

Measurements :- + Standing height + setting height + Arm span + MPC + Head circumference +wt

General Examination :-

Maneuvers:-

- **Hyperextensibility + BACK :-** Thumb, little finger, Elbow, Knee, Spine, pes plannus
- **Marfan signs:-** Thumb and wrist signs
- **Skin elasticity:-**
- **Symmetry:-**

Examination: - علي حسب الحالة

Abraam

1.Marfan

Spine and lower limbs:- بالمرة وهوا واقف وقالع الجزمة ، خليه يقلع البنطلون إن أمكن

- Café-au-lait spots
- Look from lateral for kyphosis
- Look from behind for scoliosis while standing
- Bend forward for scoliosis
- GAIT → once for any gait abnormality

Hand & Arm:- **خليه يقعد**

- Water hammer pulse
- Elbow fixed flexion or hyperextension

Head & Neck:-

- High arched palate, Dental crowding

Chest:-

- Pectus excavatum or carinatum
- Scars
- Café-au-lait spots
- I need to do Cardiac auscultation (Aortic regurgitation , Aortic root dilatation, mitral valve prolapse)
- **Gynecomastia in boys (klinefilter)**

In order to complete:-

- Cardiovascular examination including BP
- Examine parents (autosomal dominant inheritance)
- Need to plot parental height/ MPC

INVESTIGATIONS

- Bone age
- Slit lamp examination for lens dislocation
- ECHO cardiography yearly (to compare the aortic root dimensions against centile charts)
- Genetic testing for fibrillin gene on **chromosome 15**
- CT MRI for dura ectasia
- Thyroid function test to exclude hyperthyroidism as a cause for tall stature

Abraam H

MANAGEMENT

- Paediatrician (overall management , FU BP regularly , FU kyphoscoliosis and ref to orthopaedic surgeon whenever needed)
- Ophthalmologist for regular ophthalmological examination
- Cardiologist, for yearly ECHO
- Orthopedic surgeon (scoliosis)
- Occupational therapist for the hyperlaxity
- Psychiatrist

2.Hyperthyroidism

Hand:- tremors, tachycardia

Head and Neck:-

- **Eye signs of hyperthyroidism :- exophthalmos, lid signs**
- **Goiter, inspect and palpate**

Full thyroid examination

3. Homocystinuria

- **Development and fundoscopy**

4.Klinefilter

- **Examine genitalia and pubertal staging**

Abraam H

SHORT STATURE

Disproportionate

Short Arm Span

- Achondroplasia :- Rhizomelic
- Ellis Van Creveled

Normal Arm span

- MPS (Short trunk)
- Rickets (might be dispproportionate BCZ of wt bearing)
- Osteogenesis Imperfecta

Proportionate

Dysmorphic

- Noonan
- Tturner
- Russel Silver
- MPS
- Other syndromes for DD and whole genomic sequences

Non Dysmorphic

- Growth Hormone deficiency
- Chronic diseases
  - CRF
  - IBD
  - CF
  - Celiac
  - DM I
- Rickets
- Osteogenesis Imperfecta

Abram

GENERAL RULES:

1. Female in a short stature station >> you should exclude Turner
2. In any short stature station you have to examine the thyroid gland(hypothyroidism could be associated with Turner or alone).
3. Pubertal assessment is crucial
4. Rickets could be the case

GENERAL observation:- as usual + lokks short so I'd like to take his measurements

Measurements :- + Standing height + setting height + Arm span + mid parental centile

*** Turner, Down and Achondroplasia have specific growth charts.**

I Would like to do general examination :-

MANEUVERS:-

- افرد ايديك ورجليك قصاد بعض **FOR ASYMMETRY or Hemihypertrophy → Russell silver**
- افرد ايديك لقدام
  - Nails: hypoplasia, hyperconvex → turner
  - **Small trident hand → Achondroplasia**
  - **Short stuppy fingers→ (mucopolysacharidosis)**
  - **Scar of carpal tunnel synd. Operation→ (mucopolysacharidosis)**
  - **Clinodactyly → Down syndrome and russell silver**
  - **Camptodactyly (fixed flexion of fingers)→ Russell Silver**
  - Simian crease → down syndrome
- **Make a fist** → short 4th & 5th metacarpals (turner)
- **Extend elbows** → Carrying angle (turner, noonan)
- **Extend elbows → short limb , not reaching upper thigh → Achondroplasia**
- **Flex elbows → for rhisomelic shortening → Achondroplasia**
- **Flex elbows → for mesomelic shortening →Ellis van creveled**
- **Proximal myopathy, thin skin → cushing syndrme**

Back + LOWER LIMBS :- **NB:- YOU HAVE TO EXPOSE LL TO EXCLUDE RICKETS**

- **Skin → to exclude neuro cutaneous**
- **exaggerated kyphosis, gibbus, scoliosis and rod surgery → (mucopolysacharidosis)**
- **Scars of corrected deformity eg. Cervical stabilization surgery In Maroteaux-Lammy and Morquio**
- **Gibbus in infancy , lumbar lordosis and scoliosis in adolescence→ achondroplasia**

EXAMINATION According to the case :-

1. **Turner**
2. **Noonan**
3. **Russel silver**
4. **MPS**
5. **Ellis-Van Creveld syndrome // Achondroplasia**
6. **Rickets**
7. **Osteogenesis Imperfecta**
8. **Growth hormone deficiency**
9. **Chronic disease**

Abraam H

Turner Syndrome

Hand:- **Examine pulse for AS**

Head & Neck :-

TURNER

- **High arched palate**
- **Neck webbing**
- **Low posterior hair line**
- **Goiter**

Chest:-

- Shield shaped chest
- widely spaced nipples
- Absent breast tissue
- Scars

Abdomen:-

- Scars (kidney operations)

Can I do :-

- cardiac auscultation
- abdomen examination for any renal masses

In Order to complete:-

- **Pubertal tanner staging**
- Full cardiovascular examination including peripheral pulses and 4 limb BP
- Ear examination & formal hearing test (chronic serous otitis media & sensori-neural hearing deficit)
- Thyroid status(autoimmune hypothyroidism)
- Plot height, wt ,head circumference on special charts for Turner syndrome

Abraam H

TURNER

- Management:- **MDT**
  - **Geneticist** :- Confirm diagnosis by Karyotyping
  - **Cardiologist**:- ECHO
  - **Endocrinologist**:- Growth hormone replacement therapy (follow up to monitor growth & side effects) & for Oestrogen/progesterone replacement therapy
  - **Audiologist:-** for hearing assessment
  - Screen for associated autoimmune diseases like:- autoimmune thyroiditis and inflammatory bowel disease
  - Screening for renal anomalies by **abdomen US**
  - Follow up to monitor for complications including hypothyroidism, hearing loss, hypertension, scoliosis, insulin resistance, neurodevelopmental difficulties, amblyopia.

Difference between Turner and Noonan:-

Cognitive

Cardiac

Coagulopathy

Chromosomal

Abraam

NOONAN syndrome

EXAMINATION:-

Head & Neck :-

NOONAN

- **Dysmorphic features (hypertelorism, antimongoloid slant, ptosis, epicanthic fold, low set ears , short webbed neck)**

Chest:-

- Shield shaped chest
- widely spaced nipples
- Pectus
- Scars
- Heart auscultation (PVS, branch PAS, ASD, hypertrophic cardiomyopathy)

In order to complete:-

NOONAN

- Abdominal examination for hernias
- Genitalia examination for cryptorchidism
- Developmental assessment (they have learning difficulties)
- Ear examination and formal hearing test (chronic otitis media and hearing loss)
- Examine parents(autosomal dominant)

Management:-

- **INVX:-**
  - Genetic testing to confirm diagnosis chromosome 12
  - ECHO **(**screening for cardiac anomalies (Pulmonary valve stenosis, septal defects, hypertrophic cardiomyopathy)**)**
  - Coagulation profile(coagulation abnormalities in 30% eg:- Von Willebrand, thrombocytopenia, clotting factors abnormalities)
  - Hearing test

- **MDT:-**
  - Geneticist for genetic counseling
  - Speech therapist if there are speech and articulation issues
  - Physiotherapist and occupational therapist for gross and fine motor delays
  - Hematologist for coagulation problems
  - Cardiologist

Russel Silver

EXAMINATION:- pt will look cachectic

Hand:- asymmetry, syndactyly, clinodactyly, camptodatyly

Head & Neck :-

Russell silver

- **Dysmorphic features (frontal possing, normal head circumference, small triangular face, micrognathia, , prominent nasal bridge, downturned corners of mouth, late closure of AF)**
- **Blue sclera**
- **Sprengles neck deformity**

Abraam

In order to complete my examination:-

- Genitalia examination (hypospadias/ undescended testis)
- Abdominal examination (for masses to exclude Wilm's tumour)

Management:-

- **History :-** Review the child's growth chart (borne small for gestational age, little or no catch-up growth)
  - **INVX**
    - Genetic testing (all genome sequence bcz of multiple gene affections on chromosomes 7 , 11 , 15)
    - ECHO to screen for any associated cardiac anomalies
    - Abdomen US to Screen for tumours eg:- wilm's, or Renal anomalies
    - Bone age will be delayed
    - X-ray looking for the characteristic changes:-
      - 'ivory' epiphyses of the distal phalanges.
      - Small middle phalanx of little finger.
      - Pseudoepiphysis at the base of the second metacarpal.

- **MDT:-**
    - ✓ Geneticist
    - ✓ Orthopaedic surgeon (for marked limb asymmetry)
    - ✓ Dietitian
    - ✓ Occupational therapist
    - ✓ Physiotherapist
    - ✓ Endocrinologist
    - ✓ Community paediatrician

=--=-=-=-

Mucopolysacharidosis

EXAMINATION:- Submental Dysmorphic

Head & Neck :-

MUCOPOLYSACHARIDOSIS

- **Coarse facial features (thickened lips, frontal possing, large head, flat mid face, prognathism)**
- **Corneal clouding (in Hurler) (اوعي تلمس وشه، الفت انتباهه بس)**
- **Nasal discharge**
- **Upper air way obstruction**

Chest:-

- Heart auscultation (for congestive hrt failure)

Abdomen:-

- Abdominal distension
- Hernial orifices (especially umbilical hernia)
- Hepatosplenomegaly (ask permission)

Main problems

- Coarse features
- MSK (joints stiffness and contractures)
- Submentality
- hepatosplenomegaly

Back + LOWER LIMBS :- **NB:- YOU HAVE TO EXPOSE LL TO EXCLUDE RICKETS**

- **exaggerated kyphosis, gibbus, scoliosis and rod surgery → (mucopolysacharidosis)**
- **Scars of corrected deformity eg. Cervical stabilization surgery In Maroteaux-Lammy and Morquio syndrome**

Ahm H

In order to complete:-

MUCOPOLYSACHARIDOSIS

- CVS examination for evidence of congestive heart failure
- MSK examination for joints stiffness and contractures
- Developmental assessment (delayed)
- Fundus examination for cherry red spots (present in GM1,which is a differential)

Management:-

- **INVX:-**
    - Enzyme assay in leukocytes ()
    - Urine glycosaminoglycans (GAGs)
    - Geneticist →Genetic testing / counseling
    - Abdomen US
    - Cardiologist →ECHO (cardiomyopathy)
    - Spine X-Ray for kyphosis, scoliosis and Gippus
    - Audiologist →Hearing assessment
    - Ophthalmologist →vision assessment / corneal cloudening
    - ENT →Sleep studies for OSA
- **MDT:-**
    - Physiotherapist and occupational therapist(Joint stiffness)
    - Orthopaedic surgeon (kyphosis and scoliosis)

Other DD:-

- Multiple sulfatase deficiency (icthyosis, more profound neurological deterioration)
- GM1 gangliosidosis (pseudo-Hurler)
- Mannosidosis (clinically like Hurler without urinary findings)
- Fucosidosis (clinically like Hurler without urinary findings)
- Mucolipidosis

Abraam H

Achondroplasia

EXAMINATION:-

Head & Neck :-

ACHONDROPLASIA

- **Dysmorphic features :- (frontal possing, large head apparently only, flat nasal bridge)**
- **High arched palate**
- **Teeth malocclusion**

Back + LOWER LIMBS :- **NB:- YOU HAVE TO EXPOSE LL TO EXCLUDE RICKETS**

- **Gibbus in infancy , lumbar lordosis and scoliosis in adolescence→ achondroplasia**

In order to complete:-

ACHONDROPLASIA

- Spirometry → bcz all pts have reduced lung capacity
- CVS examination for evidence of pulmonary HTN 2ndry to recurrent sleep apneas
- Ears examination for chronic serous otitis media
- MSK examination for ligaments hyperlaxity
- HC → can have hydrocephalus
- Plot HC, HT & WT on special achondroplasia charts (hydrocephalus)
- Examine parents (autosomal dominant inheritance)

Management:-

- **INVX:-**
    - Genetic testing (AD 4p, 50% neomutation)
    - ECHO to check for pulm HTN
    - X-ray spine:- to check for decreasing interpeduncular distances in lumbar spine. (distinguishes it from Hypochondrodysplasia)
    - Audiologist → Hearing assessment
    - MRI to exclude coning or herniation
    - ENT →Sleep studies for OSA

- **MDT:-**
  - Geneticist for genetic counseling (AD 4p)
  - Orthopaedic surgeon (gibbus , scoliosis)
  - Cardiologist for pulm HTN
  - Pulmonoligist for management of OSAs
  - Physiotherapist and occupational therapist
  - Community paediatrician
  - Dentist for dental malocclusion

Recurrence :-

If no family history it's 1-3%

If there is family history → 50%

Main cause of death :- cervical cord compression because of small foramin magnum

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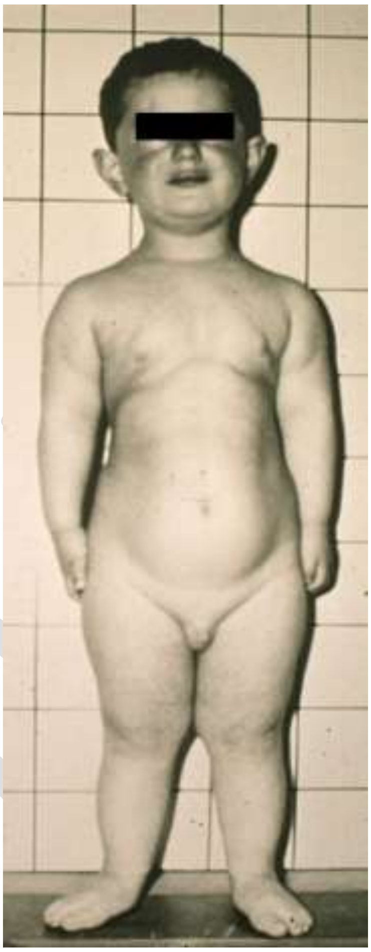
Abraam H

Ellis-Van Creveld syndrome



HAND:-

- Poly dactyly (postaxial)
- Finger nail dysplasia
- Mesomelic shortening

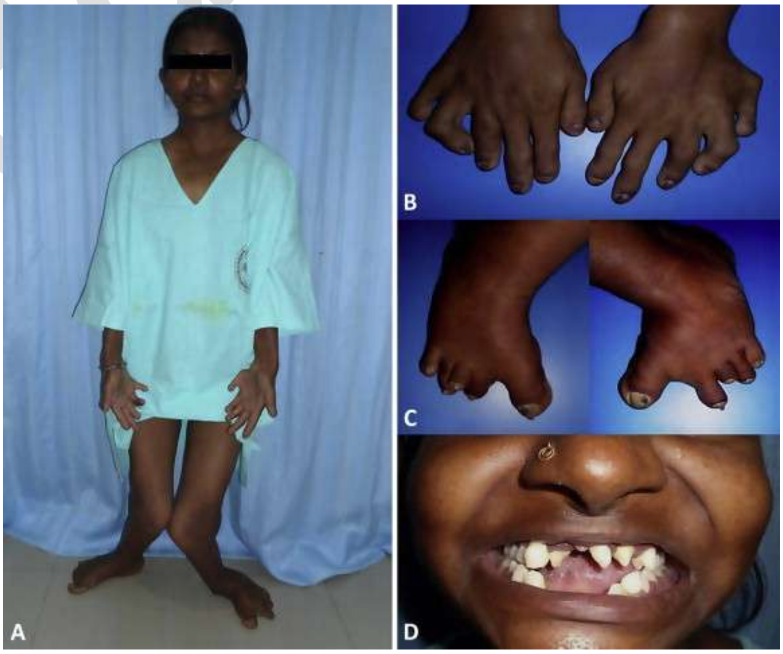
Head and Neck:-

- Natal teeth
- Abnormal teeth
- Cleft palate

Chest:-

- Heart defects (murmur)
- Scar of surgery
- Short ribs

Lower limb:- Deformity (genu valgum) extra toe



In order to complete my examination:-

- Full CVS examination
- Developmental assessment

Management:-

- **INVX:-**
  - Genetic testing (4p)
  - ECHO
  - Hand X-Ray
- **MDT:-**
  - Geneticist (for genetic counseling ,, AR)
  - Cardiologist
  - Dentist

Abraam

Rickets

Hands:-

- Broadening of wrist and elbow joints
- Hypermobility of the Thumb, little finger and elbow.
- Obvious limb deformities from previous fractures.
- Tanner staging at the axilla if at puberty

Head & Neck:-

- Shape
    - Frontal bossing
    - Box shaped head
    - Alopecia totalis → in Vitamin D dependent rickets type 2
- Size → Measure the head circumference (فك الضفيرة !!)
- Sutures → any prominent sutures
- Scars → any scars
- Mouth → teeth

Chest:-

- Rosary beads (ارفع ايدك فوق راسك وميل لناحية)
- Hrrison's sulci
- pigeon shaped chest (pectus carinatum)

Abdomen:-

- examine the liver → Hepatic rickets

Lower limb:-

- genu varum / valgum
- marfan sign

Differential diagnosis:-

1. The obvious reason after examination :-
    a. Hypophosphatemic rickets
    b. Vitamin D dependent rickets type 2 → Alopecia totalis

Abraam H

2. Hepatic rickets
3. Renal rickets
4. The other Types:-

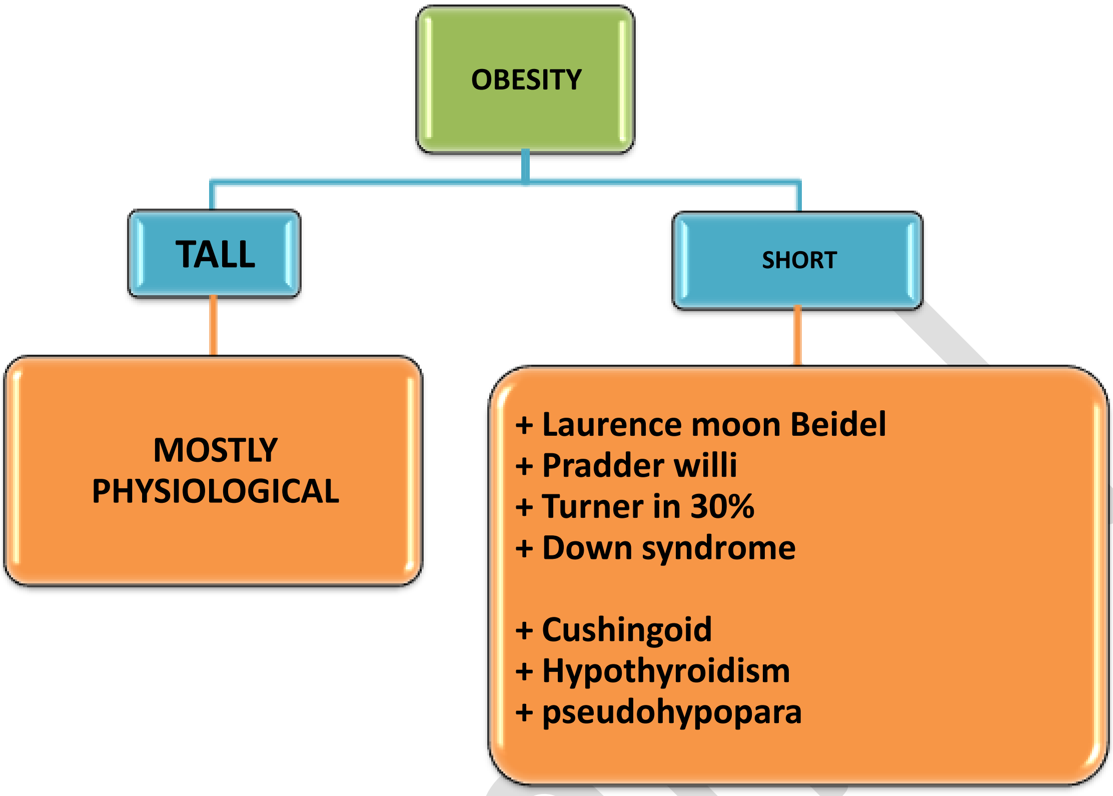
    a. **Vitamin D-related rickets**
        i. **Vitamin D deficiency**
            1. Nutritional→(the commonest cause)→ poor sun exposure, poor feeding.
            2. GIT loss:- celiac, cystic fibrosis, steatorrhea (poor absorbtion)
        ii. **Vitamin D-dependent rickets**
            1. Type 1 (vit D 1-alpha-hydroxylase deficiency)→ active vit D deficiency
            2. Type 2 (calcitriol receptor mutation)→ receptor problem
    b. **Hypocalcemia-related rickets**
        i. Hypocalcemia
        ii. **Chronic renal failure** → renalosteodystrophy (dec Ca, incr PTH → Bone degradation
    c. **Hypophosphatemia-related rickets**
        i. **Congenital**
            1. Vitamin D-resistant rickets (X- linked hypophosphatemic rickets) → decreased Ph reabsorbtion + decreased vit D synthesis, so TTT → oral Ph + Calcitriol
            2. Autosomal dominant hypophosphatemic rickets(ADHR)
            3. Autosomal recessive hypophosphatemic rickets (ARHR)
        ii. **Secondary Hypophosphatemia** (typically secondary to malabsorption)
        iii. **Fanconi's syndrome** → ph loss from the proximal tubules

MANAGEMENT:-

1. I would like firstly to take a thorough history especially nutritional history & family H.
2. Investigations will include :-
    a. bone profile in the form of Ca, Ph, ALP &vit. D
    b. x-ray of lower ends of long bones>> widening epiphyseal plates, cupping and fraying together with increased joint space.
    c. Genetic testing for inherited reasons
3. Finally I would like to involve a **MDT**:

- Occupational therapist, My consultant, Orthopedic(in severe cases), Health visitor, Radiologist(green sticks are common), Endocrinologist, Geneticist for counselling

OBESITY



GENERAL Observation:- as usual+ he looks OBESE , so I'd like to plot his height and weight on appropriate centile charts for age and sex, also I'd like plot his **BMI ,and know his parents' BMI**

Measurements :- + height +weight **+BMI**

BMI:-

- ✓ FROM 91- 98% overweight
- ✓ From 98-99.6 % obese
- ✓ Above 99.6% Morbid obese

Laurence moon beidel → obese, wearing glasses, submental

Prader willi → obese, Narrow forehead, may be normal IQ

Examination :-

Gait Once :- for any abnormal gait

Abraam

Back:-

- Interscapular pad of fat
- Kyphosis & scoliosis

Hand & Arm:-

- Short fingers (Prader willi)
- Scar in the hand of polydactyly removal (Lawrence moon Beidel)
- Short 4th and 5th metacarpals (turner)
- Muscle wasting, thin skin (cushingoid)
- BP

Axilla:-

- Acanthosis nigricans
- Tanner staging

Side of the neck:-

- Webbing
- Acanthosis nigricans

Head:-

- Bifrontal narrowing (prader willi)
- Glasses → Lawrence moon beidle
- Eyes for cataract → cushing
- Moon face → Cushing
- Facial hair, Acne → Cushing

Mouth:-

- Thrush of stomatitis →cushing
- High arched palate → turner

Neck:-

- Goiter

Chest:-

- **Inspect:-**
    - Truncal obesity

Abraam H

  - Gynecomastia
  - Signs of turner → scars , widely spaced nipples , shield shaped chest
- **Auscultate:-**
  - Chest → wheezes
  - Heart → accentuated P2 (obstructive sleep apneas) + any murmur (in turner)

Abdomen:-

- **Inspection:-**
  - Striae
  - Scars (adrenal surgery , GIT disease requiring steroids)
- **Palpation :-**
  - Renal or suprarenal masses
  - Hepatomegaly → fatty liver

Offer Genitalia:-

- Hypogonadism in pradder willi
- Precoscious puperty in Cushing

Lower limbs:-

- Myopathy in Cushing
- Pedal myxedema in hypothyroidism

In order to complete:-

- **Measure BP**
- **Tanner staging**
- Fundoscopy and visual field examination (pituitary tumours)
- Full systems examination to check for any disease requiring exogenous steroid treatment

DD:-

- Physiological
- Pathological
  - Chromosomal/Genetic
    - Prader-Willi
    - Lawrence –Moon-Beidel
    - Beckwith-Wiedemann
    - Down

Abraam H

- Endocrine
    - Cushing Syndrome
        - Primary
            - Adrenocortical tumor
            - mcCune Albright syndrome
        - secondary
            - exogenous steroids
            - ACTH secreting pituitary tumor
            - ACTH secreting Ectopic tumor
    - Hypothyroidism
    - Growth hormone deficiency
    - Polycystic ovary syndrome

Management:- take thorough history

- **INVX:-**
    - Identification of underlying cause:-
        - Thyroid function tests
        - 24h urinary cortisol and plasma cortisol measurements at midnight and 8am as a screening test when cushings syndrome is suspected
        - ACTH level
        - Dexamethasone suppression test
        - CT head/abd :- screening for tumours (adrenal tumour, pituitary tumour) / wilms tumour
    - Screening for associations and complications
        - Insulin level
        - blood sugar / oral glucose tolerance test
        - lipid profile
    - Bone age
- **MDT:-**
    - Sleep studies (ENT specialist)
    - **Dietician**
    - **Endocrinologist**
    - Cardiologist →ECHO (Pulmonary HTN)
    - Psychologist
    - Orthopaedic surgeon for any orthopaedic problems

Akram H

Others General examination

Gait Once :- for any abnormal gait

Back:-

- Interscapular pad of fat
- scars
- Kyphosis & scoliosis

Hand & Arm:-

- Fingers
- Hands
- Arms
- Axilla

Side of the neck:-

- Webbing
- Acanthosis nigricans

Head:-

- Any abnormal features:-
- Hair line
- Forehead
- Eyes :-
  - Ptosis
  - Hepertelorism
  - Palbebral fissure
  - Corneal abnormality
  - Cataract
  - Microphthalmia
  - synophrys
- Nasal bridge
- Nose peaked?
- Philtrum long?
- Mouth:-
  - High arched palate → حاول تلمس سقف حلقك ب لسانك

Abraam H

  - Tongue
  - Teeth
- Mandible
- Ears :-
  - Low set
  - Microtia

Neck:-

- Goiter :-
  - Stick your tongue out
  - swallow

Chest:-

- **Inspect:-**
- **Auscultate:-**
  - Chest →
  - Heart → Heart sounds + any murmur

Abdomen:-

- **Inspection:-**
- **Palpation :-** Organs and masses

Offer Genitalia:-

Lower limbs:-

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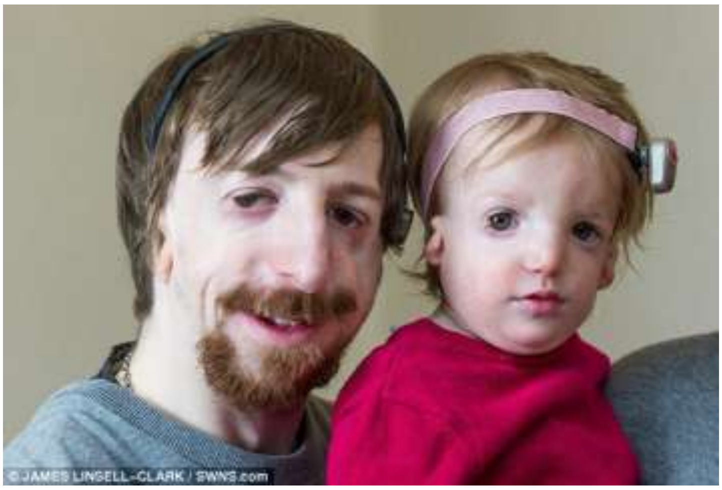
Some syndromes

Tracher Collins syndrome :- AD


© JAMES LINSELL-CLARK / SWNS.com

- Hypoplastic maxilla
- Downslanting palbebral fissure
- Cloboma in eye lid
- Microtia
- Cleft palate
- Hearing difficulties

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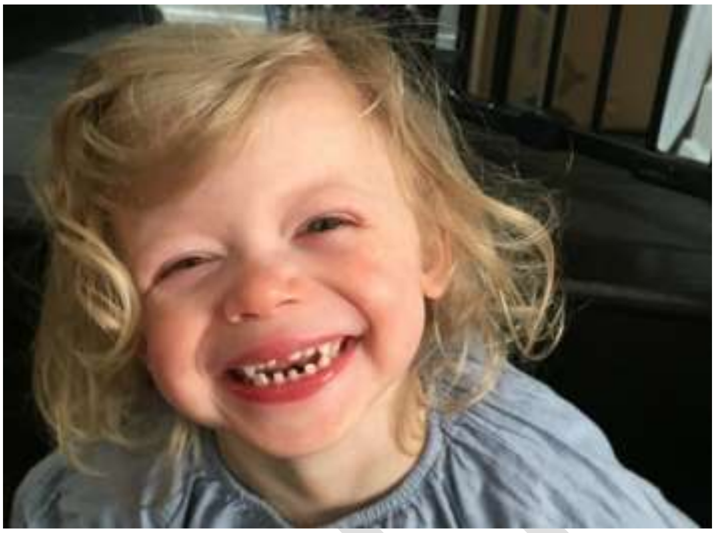
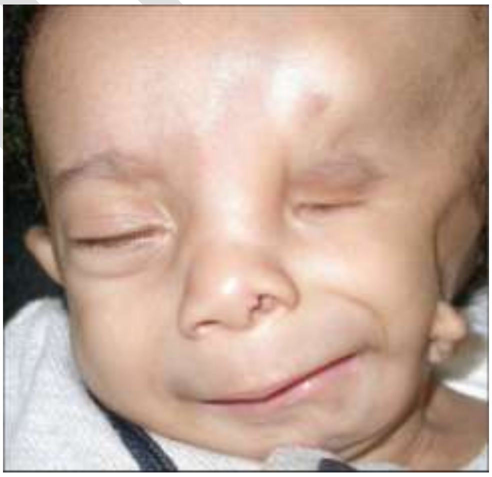
Williams syndrome:-



- Microdeletion on chromosome 7
- Short stature
- Characteristic facies
    - Broad forehead
    - Short peaked nose
    - Long philtrum
    - Ear to ear smile
    - Full cheeks
    - Puffy eye lids
- Cocktail party character
- Mild to moderate learning difficulty
- Examine heart & BP for supravalvular AS + Kidneys
- Infantile hypercalcemia

Goldenhar syndrome:-



- Facial asymmetry
- Half of the face grows normally,
  but the other half is static causing:-
    - Microtia
    - Micophthalmia
    - Deviated chin towards the static half

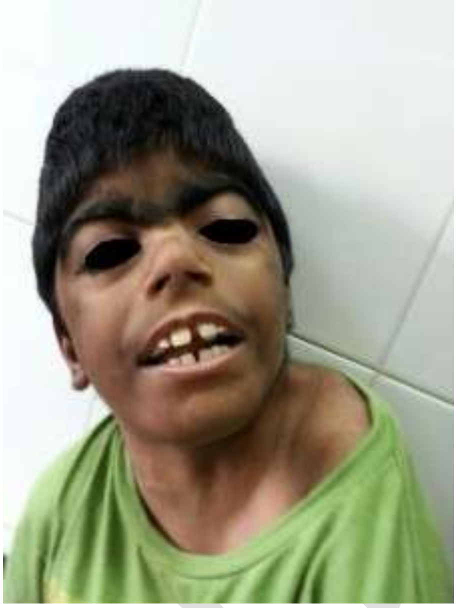
Ehlers Danlos syndrome:-

Ehlers–Danlos syndromes (EDS) are a group of genetic connective tissue disorders. Symptoms may include loose joints, stretchy skin, and abnormal scar formation. These can be noticed at birth or in early childhood. Complications may include aortic dissection, joint dislocations, scoliosis, chronic pain, or early osteoarthritis.

There is no known cure. Treatment is supportive in nature. Physical therapy and bracing may help strengthen muscles and support joints. While some types result in a normal life expectancy, those that affect blood vessels generally result in a shorter life expectancy.

Abraam N

Cornelia De Lange Syndrome:

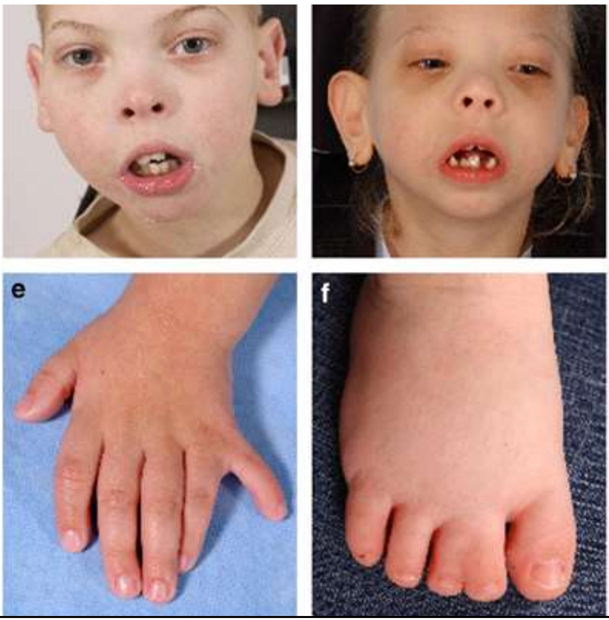


- Microcephaly
- Low hair line
- Synophrys
- Long eye lashes
- Hypertelorism
- Depressed nasal bridge
- Long philtrum
- Abnormal teeth
- Absent digits
- Associated cardiac anomalies
- Global Developmental delay

Smith-Lemli-Opitz syndrome :-

is a developmental disorder that affects many parts of the body. This condition is characterized by distinctive facial features, small head size (**microcephaly**), **intellectual disability or learning problems**, and behavioral problems. Many affected children have the characteristic features of **autism**, a developmental condition that affects communication and social interaction. Malformations of the heart, lungs, kidneys, gastrointestinal tract, and genitalia are also common. Infants with Smith-Lemli-Opitz syndrome have **hypotonia**, experience **feeding difficulties**, and tend to grow more slowly than other infants. Most affected individuals have fused second and third toes (**syndactyly**), and some have **polydactyly**.

The signs and symptoms of Smith-Lemli-Opitz syndrome vary widely. Mildly affected individuals may have only minor physical abnormalities with learning and behavioral problems. Severe cases can be life-threatening and involve profound intellectual disability and major physical abnormalities



Fragile X syndrome:-

(FXS) is a genetic disorder. Symptoms often include **mild to moderate intellectual disability**. Physical features may include a **long and narrow face**, **large ears**, **flexible fingers**, and **large testicles**. About a third of people have features of **autism** such as problems with social interactions and delayed speech. **Hyperactivity** is common and **seizures** occur in about 10%. Males are usually more affected than females.

Fragile X syndrome is typically due to an expansion of the CGG triplet repeat within the Fragile X mental retardation 1(FMR1) gene on the X chromosome. This results in not enough fragile X mental retardation protein (FMRP), which is required for normal development of the connection between neurons. Diagnosis is by genetic testing to determine the number of CGG repeats in the FMRI gene. Normal is between 5 and 40 repeats, fragile X syndrome occurs with more than 200, and a premutation is said to be present when a middle number of repeats occur. Testing for premutation carriers may allow for genetic counseling

There is no cure. Early intervention is recommended as it provides the most opportunity for developing a full range of skills.[4] These interventions may include **special education, speech therapy, physical therapy, or behavioral therapy.** Medications may be used to treat associated seizures, mood problems, or ADHD. Fragile X syndrome occurs in about 1 in 4,000 males and 1 in 8,000 females.

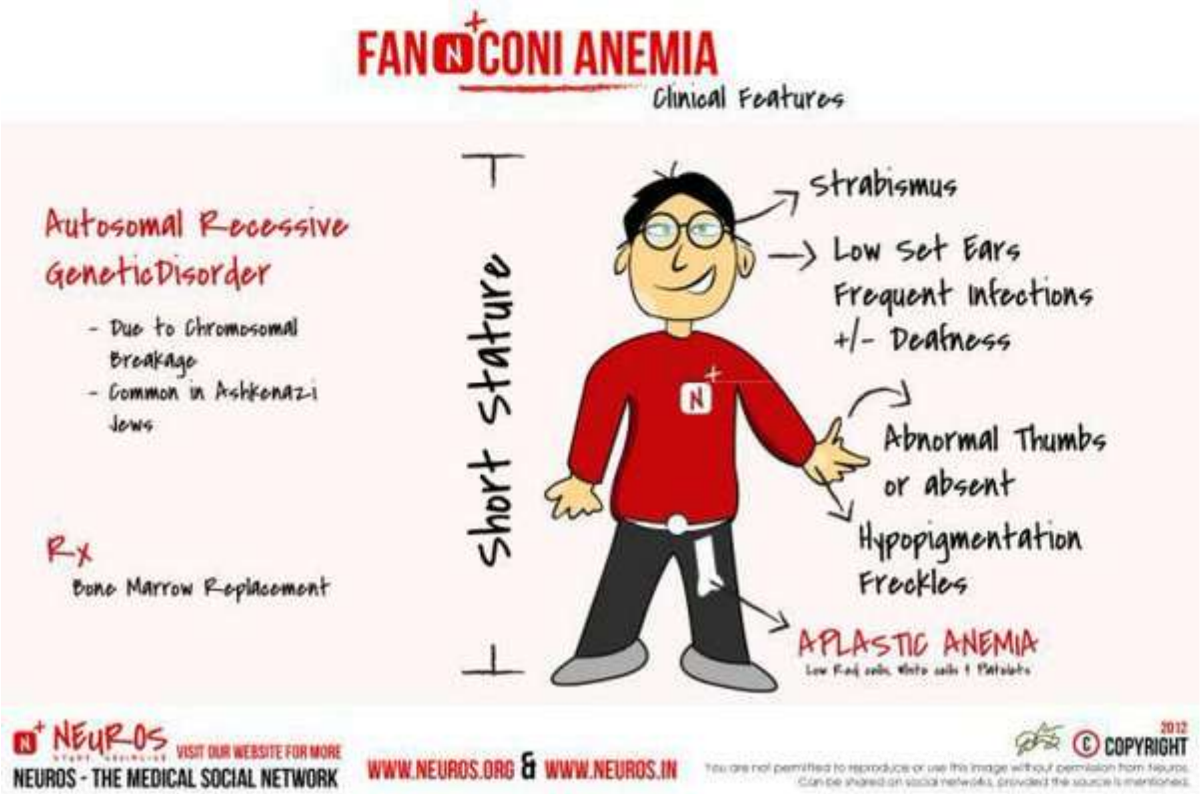
Abraa

Fanconi anemia

Fanconi anaemia (FA) is a rare genetic disease resulting in impaired response to DNA damage. Among those affected, the majority develops **cancer**, most often acute myelogenous **leukemia**, and 90% develop **bone marrow failure** by age 40. About 60–75% of people have congenital defects, commonly **short stature**, **abnormalities of the skin, arms, head, eyes, kidneys, and ears, and developmental disabilities**. Around 75% of people have some form of endocrine problems, with varying degrees of severity.

FA is the result of a genetic defect in a cluster of proteins responsible for DNA repair.

Treatment with androgens and hematopoietic growth factors can help bone marrow failure temporarily, but the long-term treatment is bone marrow transplant if a donor is available.


FANCONI ANEMIA
Clinical Features
Autosomal Recessive GeneticDisorder
- Due to Chromosomal Breakage
- Common in Ashkenazi Jews
Rx
Bone Marrow Replacement
Short stature
Strabismus
Low Set Ears
Frequent infections
+/- Deafness
Abnormal Thumbs or absent
Hypopigmentation
Freckles
APLASTIC ANEMIA
Low Red cells, White cells & Platelets
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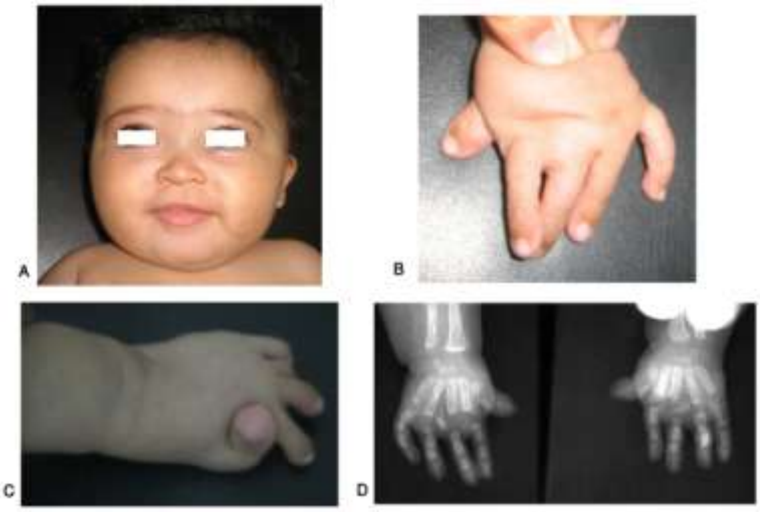
Rubinstein–Taybi syndrome:-

Rubinstein–Taybi syndrome (RTS), also known as broad thumb-hallux syndrome or Rubinstein syndrome, is a condition characterized by **short stature**, moderate to severe **learning difficulties**, **distinctive facial features**, and **broad thumbs and first toes**. Other features of the disorder vary among affected individuals.

People with this condition have an increased risk of developing noncancerous and cancerous tumors, leukemia, and lymphoma. This condition is sometimes inherited as an autosomal dominant pattern and is uncommon, many times it occurs as a de novo



Crouzon, Apert , Pfeiffer syndrome (cranio-facial-dysostosis)

https://en.wikipedia.org/wiki/Crouzon_syndrome

https://en.wikipedia.org/wiki/Apert_syndrome

https://en.wikipedia.org/wiki/Pfeiffer_syndrome

Neurofibromatosis

General observation:- Large Head, , neurofibromas, café au lait,

Examination:- Justified exposure

Gait & Back :- for café au lait spots + Scoliosis → Count and measure

Common :-

= Neurofibromata :- painfull or not?? average number? size and shape, attached to the underlying skin??

= Café au lait spots :- colour, average number, border, the size of the largest one.

- **I suspect neurofibromatosisi and I'd like to examine for other signs of neurofibromatosis.**

Upper limb:-

- Axillary freckling
- Asymmetry

Head:-

- Large head (measure)
- Palpate for any shunt (ابتدي حس من الرقبة)
- Eye
- Sphenoid dysplasia

Trunk:-

- Café au lait spots :- colour, average number, border, the size of the largest.
- Abdomen for scar of the shunt

Lower Limbs:-

- Deformity :- bowing
- Asymmetry

Abraam H

Back:-

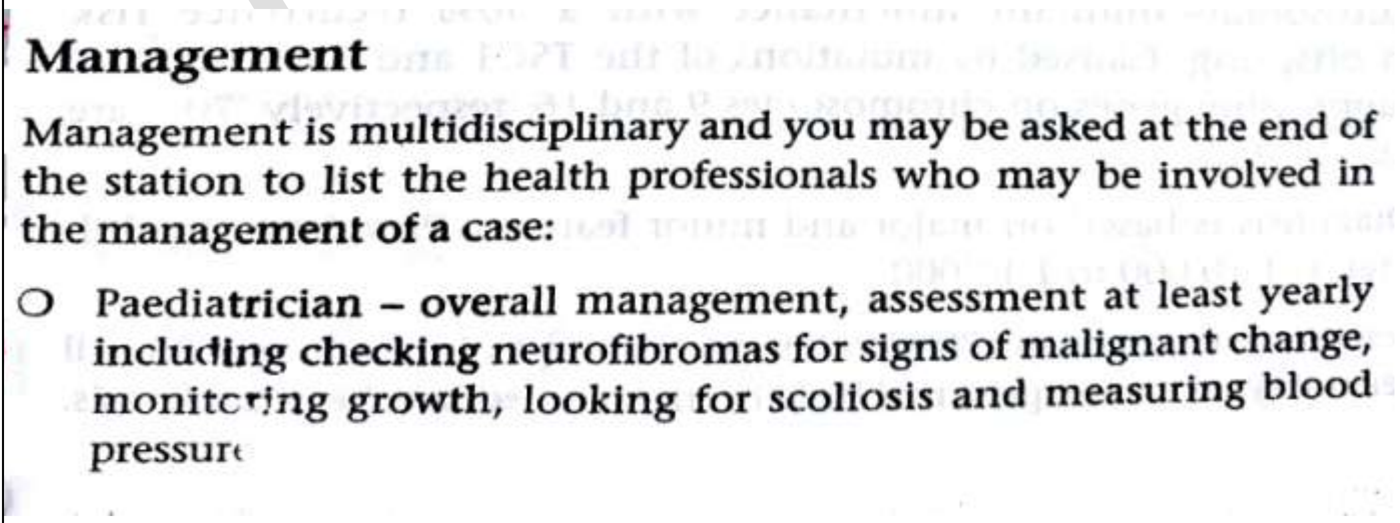
- Kyphosis
- Scoliosis

In Order to complete:-

- BP (pheochromocytoma and renal artery stenosis)
- Complete CVS examination including :- Femoral pulsations (coarctatation)
- Abdominal examination (wilm's tumour)
- Ophthalmological examination :- visual acuity (for optic glioma)& slit lamb for lisch nodules,
- Inguinal freckling
- Growth parameters (short stature)
- Examine parents for autosomal dominant inheritance
- Developmental assessment → 1/3 has learning disability

Management:-

1. History , genetic counseling and pedigree, FH ….
2. Investiagations :-
    a. Slit lamb eye examination
    b. **Bloods:-** tumour markers, urinary catecholamines, GH levels
    c. **Radiological INVX**:- ECHO, CT brain and Abdomen , Renal Doppler(RAS), spine X-ray
    d. **Screening for the possible associated tumours**:- optic glioma, astrocytoma, meningioma, ependymoma, phaeochromocytoma, Wilm's tumour, Thyroid carcinoma.
3. **MDT:**
    4. → mark beattie page 351

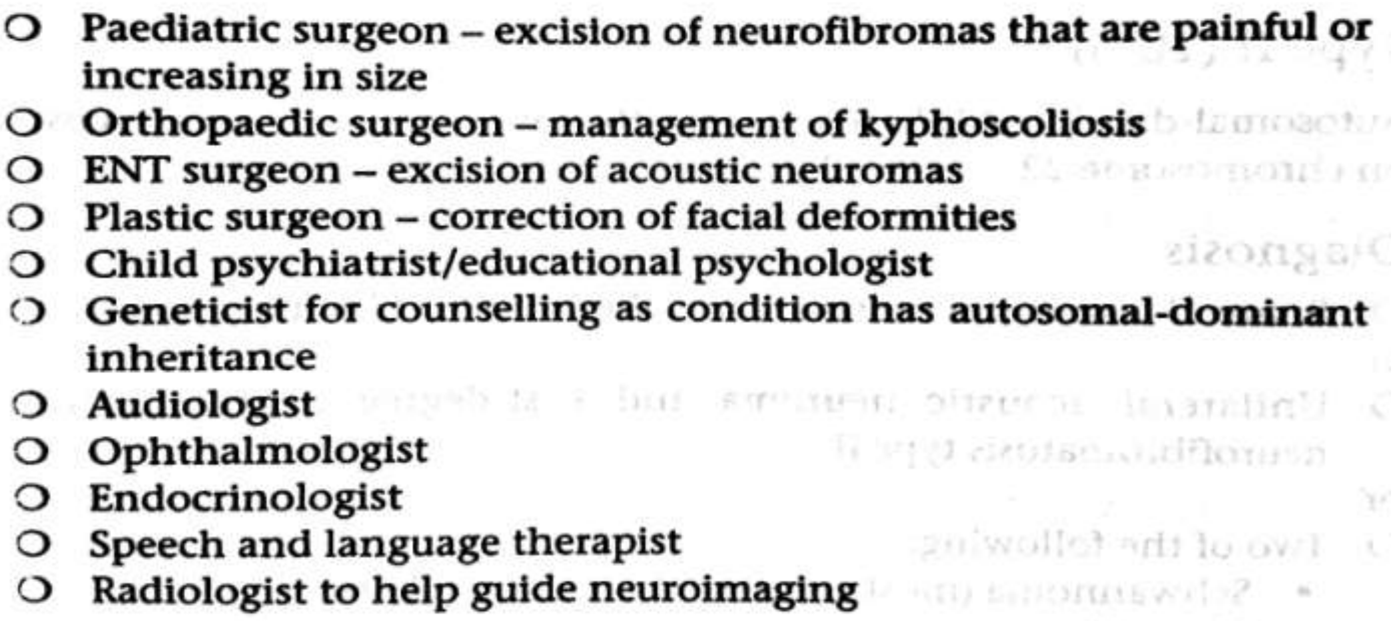


Management

Management is multidisciplinary and you may be asked at the end of the station to list the health professionals who may be involved in the management of a case:

○ Paediatrician – overall management, assessment at least yearly including checking neurofibromas for signs of malignant change, monitoring growth, looking for scoliosis and measuring blood pressur

Abraam H



- Paediatric surgeon – excision of neurofibromas that are painful or increasing in size
- Orthopaedic surgeon – management of kyphoscoliosis
- ENT surgeon – excision of acoustic neuromas
- Plastic surgeon – correction of facial deformities
- Child psychiatrist/educational psychologist
- Geneticist for counselling as condition has autosomal-dominant inheritance
- Audiologist
- Ophthalmologist
- Endocrinologist
- Speech and language therapist
- Radiologist to help guide neuroimaging

N.B. causes of hypertention in NF1:

- Pheochromocytoma
- RAS
- Coarctation
- Noradrenaline coming from the neurofibromas

Recurrence ? 50% if FH +ve , 1-3 % if FH –ve

Diagnosis? 2 major criteria out of 7

Advice:- FU yearly for BP, Neurofibromas, eyes, Bones, Brain imaging

Surgery :- might be needed for removal of neurofibromas, may be for scoliosis, may be for Renal angiomyolipomas

Height:- he might gain height if still before puberty

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Tuberous Sclerosis

General observation:- as usual+ obvious warty lesions (adenoma sebaceum), learning disability

Examination:- Justified exposure

Gait & Back :- café au lait spots, ash leaf maccules, shagreen patch in the lower back , Exclude scoliosis and kyphosis

Hand :-

Abraam

- Periangual fibroma
- Cafe au lait spots
- Axillary freckling

Head/Neck:-

- Large Head (hydrocephalus, may be shunt)
- Adenoma sebaceum
- Eyes (any glasses)
- Enamel hypoplasia

Chest:-

- Cafe au lait spots
- Ash leaf macules

Abdomen:-

- Any scars → renal operations
- Cafe au lait spots
- Ash leaf macules

In order to complete:-

- Full eye examination (Acuity, Field, Movement, accommodation) including fundoscopy for choroidal hamartomas and Retinal Phakomas
- Rectal examination for polyps
- BP
- Abdomen examination for any masses, polycystic kidneys
- CVS examination →Rhabdomyoma
- I'D like to ask the mother about:-
    - History of seizures, any antiepileptic medications
    - Any previous imaging results
    - Any problems with vision

Management:-

- Thorough history+ FH (AD inheritance chromosomes 9&16)
- **INVX:-**
    - Examine skin by wood's light for ash leaf maccules
    - Imaging:-

Abraam H

        - Renal US:- polycystic kidneys, Renal angiomas
        - CT / MRI brain:- for tumours (subependymal giant cell astrocytoma)
        - EEG:- epilepsy
        - ECG:- WPW
        - Echocardiography:- Rhambdomyomas
- **Multidisciplinary team:-**
    - Paediatrician :- for FU and overall management
    - Cardiologist:- Rhabdomyoma
    - Geneticist:- for genetic counseling
    - Paed neurologist:- for FU and management and antiepileptics
    - Educational psychologist → (Autism) and for neurodevelopmental FU
    - SALT for learning difficulties

Advice:- annual screening for brain tumours, Renal US for tumours , BP

Cause of abdominal pain:- tumors, RAS from angiomyolipoma

Antiepileptics:- vigabatrin

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Sturge Weber Syndrome

= General observation:- Port wine stain + learning difficulties

= Neurological examination → contralateral Hemiplegia

= Cranial nerves examination → may be multiple affection

= in order to complete:- eye examination

= Management:-

= INVX:- intraocular pressure + MRA + EEG

= MDT:-

- Neurologist → anticonvulsants
- Ophthalmologist → to manage Glaucoma
- Cosmetic ttt of the port wine stain → pulsed laser
- Hemispherectomy/Lobectomy for intractable seizures

Abraam H

DEVELOPMENT

GROSS MOTOR

WALK:- امشي

- Forward→ 1
- Backward→ 1.5
- Side→ 3
- Tip toe walking→ 3
- Forward Tandem→ 4
- Backward Tandem→ 5

Tip Toe :-

- Standing → 2Y
- Walking → 3 Y
- Running → 4y

RUN:- اجري

- Runs stiffly→ 2
- Runs smoothly (fast) →3
- Tip toe running→ 4

JUMP:- نط

- Jumps (in place) → 2
- Jump Forward →3
- Hops (by one leg in place)→ 4
- Skips (by one leg forward)→5

STAND:- اقف

- Tip toe standing→2
- One Foot:-
  - 1 sec → 3Y
  - 5 sec → 4y
  - 10 Sec→ 5Y

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B) Give him a Ball:

- Kicks a ball (standing) → 2
- Kicks a ball (running) → 4
- Throws a ball (from front of his chest)→ 2
- Throws a ball (from above the head) →2.5
- Catches a ball:
  - Front out stretched arm→ 3
  - Up out stretched arm→4
  - Near to the body (front of the chest بياخدها ف حضنه) →5
- Bounces and catches a ball→ 5

C) Ask him to climb stairs:

- Up with assistance →1.5
- UP in child manner →2
- **Up in adult manner →3**
- **Down in child manner →3**
- Down in adult manner →4

D) Ask him to ride a tricycle:

- Rides Tricycle →3

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FINE MOTOR & VISION

Cubes Crayon sCissor Beads Board Book

Clean the table, use the MUM

General observation:-

- Glasses , Hearing Aids, Walking Aids
- Vision, Hearing, Obvious squint, Nystagmus, Dysmorphic features

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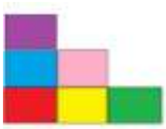
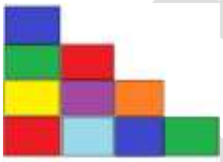
- Speech , stranger anxiety

Cubes:-

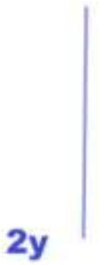
- **TOWER:-**
  - 3 cubes → 1.5 y
  - 6 cubes (age in years × 3)→ 2y
  - 9 cubes (age in years × 3) →3y
  - 12 cubes (age in years × 3)→ 4y
- **Bridge:- → 3 y**
- **Train :- → 3y**
- **Stairs :-**
  - 6 cubes → 4y

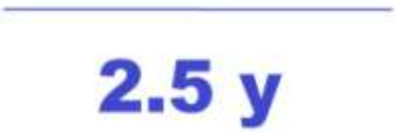


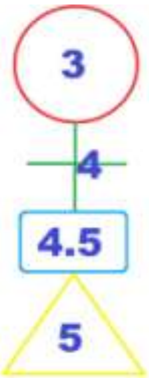
  - 10 cubes → 5y



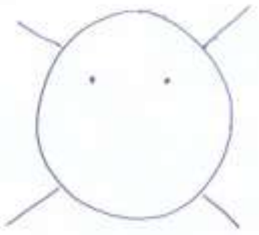
Crayon :- scribbling → 1.5



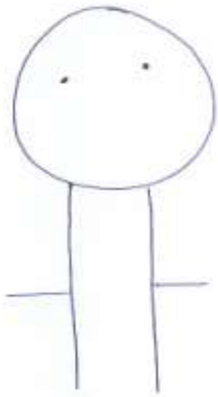




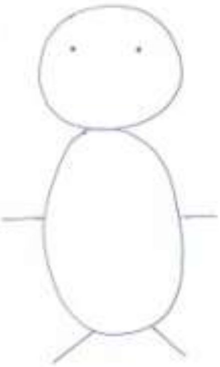
- تعرف ترسم ماما ؟



3y (Draw a man with 3 parts)



4y (Draw a man with 4 parts)



5y (Draw a man with 6 parts)

Scissors:- cut → 3 y

Beads:-

- Large hole → 3 y
- Small hole → 4 y

Board:-

- 3 shapes → 3y
- 6 shapes → 4y

Book:-

- Turn many pages → 1 y
- Turn one page → 2 y

If suspected less than 2 yrs :-

- Fix and follow
- Palmer grasp
- Transfer from hand to hand
- Mouthing

6 M

- Pointing by index
- Banging
- Crude pincer grasp
- Book → turn many pages

9 – 12 M

- Fine pincer grasp
- Tower of three cubes
- Scribbling with crayon
- Book → turn many pages

18 M

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Summary:

Folan is 4 yrs old child , well child , he was cooperative today, he showed that he could do :- …….. but he couldn't demonstrate doing:- ………… , so my impression that his fine motor age is … yrs

In order to complete my examination:- I'd like to assess other areas of development to know whether it's isolated or global developmental delay, and I'd like to send him to Child Development Center for detailed developmental assessment. And formal vision testing.

Hearing & speech

Hearing CV S C O P

Please assess child's language, Hearing, Speech

1. اقف وراه وانده عليه ------- 1سنة
2. اسمك ايه ؟ --------------- 2 سنة name
3. اسم بابا ايه ؟ -------------- 3 سنين full name
4. عندك كام سنة ؟ ------------ 3 سنين Age
5. انت ولد ولا بنت ؟ ---------- 3 سنين Sex
6. ساكن فين ؟ ---------------- 4 سنين Adress
7. تاريخ ميلادك ايه؟ --------- 5 سنين

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Sentence:-

- 2 words → 2 Y
- 3 words → 3 Y
- 4 words → 4 Y

Song:- Nursery Rhymes → 3Y

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Story:- 4 Y

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Colour recognition:-

- Color recognition, 3 colors → 4y

Counting: - باستخدام العداد أو صوابع ايديه

- Count to 10 → 4 Y

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Order command :- خد الكتاب اديه ل ماما و هات منها القلم

- Obey **simple** commands → 1.5 y
- Obey **double** commands → 2y
- Obey **triple** commands → 3y

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Parts of the Body:-

- Show 3 parts of the body → 1.5 y

Preposition :- → 3y

Pronouns:- (الملكية)→4 y

oPposite :- → 5Y الفيل و النملة ، الصيف و الشتا

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Summary:-

Folan …. Yrs old , he is doing well and cooperative child , on examination he could tell …. But he couldn't tell….. so my impression his hearing and speech age is ….

In order to complete my examination (If the assessed age is below 6 months → I'd like to do Hearing distraction test, if Below 1.5 years → Speech discrimination test)

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- And I'd like to do formal hearing test
- And I'd like to assess other areas of development to know whether it's isolated or global developmental delay.

Management :-

- Hearing aids
- Special needs school
- Speech therapist

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SOCIAL

Assess this child's play, or social development.

General observation:- responding ? , eye contact?

أكل و لبس و لعب و حمام هيستوري

A) Feeding (by examination):

- Cup → 1.5 Y

- Spoon → 1.5 Y

- Fork → 2.5 Y

- Knife → 3 Y

- Fork and Knife → 4Y

B) Dressing (by examination):

- خلع
  - Shoes → 1.5 Y
  - Shirt → 2.5 Y
- لبس
  - Dress with supervision → 3y
  - Dress without supervision → 4 Y
- زراير
  - Unbuttoning → 4Y

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    - Buttoning → 5Y

C) Play (by examination):

- **9 M →**Bye-bye , Peek a boo
- **1.5 Y →**
    - **S**ympolic play:- يللا نأكل العروسة ، يللا نسرح للعروسة (الوحيد اللي بيتعمل)
    - **S**olitary play :- بيلعب لوحده
    - **S**pectator play :- متفرج بس
- **2.5 Y →**
    - **P**retend play :- مشي المكعب كأنه عربية (يللا نمشي العربية ، بيييب)
    - **I**maginary play:- التليفون (يللا نكلم بابا)
- **3.5 Y →(by Hx)**
    - Not sharing toys < 3.5 Y
    - Sharing toys > 3.5 Y
- Play:- Group play & Role play (بيلعب استغماية و عسكر و حرامية؟) → 5 Y

D) Toileting (by history only):

- **1.5 Y →** Indicate needs
- **2.5 Y →** Dry by day
- **3.5 Y→** Go to toilet alone
- **4.5 Y→** Dry day and night

E) History :-

- Hand washing → 3Y
- Teeth brushing → 4Y
- اسأل ع اللبس لو ما عملوش

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AUTISM

- CAN BE DOWN SYNDROME
- do not disturb the child if not responding to your entrance and to your questions,(consider hearing problem)
- بيلعب لوحده
- حاول تلعب معاه
- حاول تبدل اللعبة اللي معاه ب لعبة تانية
- قول لمامته تحاول تاخد لعبته منه ، بس لو هيعيط بلاش
- أشاور علي حاجة واشوفه هينتبه وللا لأ و هيشاور وللا لأ
- أخبط ايدي و اعمل انها وجعاني و اشوفه هيتفاعل معايا وللا لأ
- After that say>> I feel that Ahmed is not responding to me, no eye contact, can I ask the mother some questions?
- Hearing?? هل حماده عنده مشكله ف السمع يا ماما ؟
- Is he speaking with you? هل بيتكلم معاكي؟
- If he wants anything?>>pointing or screaming? لما بيعوز حاجة بيشاور عليها وللا بيصرخ ويعيط؟
- Routines and repetitive behaviors? بيحب يكرر لعبه و حركاته؟
- Happy with a gift or indifferent? هل لما بتديله لعبه او هدية بينبسط بيها وللا مش بيفرق معاه؟
- الأكل :-
    - كوباية ، معلقة، شوكة ، شوكة وسكينة
- اللبس (زي فوق)
- اللعب (زي فوق) :- هل بيلعب مع اصحابه و للا علي طول بيلعب لوحده؟
- الحمام (زي فوق)

How to diagnose Autism:-

Clinical diagnosis by specific score :- Children Autism Rating Score

Red Flag signs for Autism at age of 1 year:-

- No gestures (Mask face)
- No social smile
- No babbling
- No bye bye
- No pointing
- Regression (loss of acquired language or skills)

Management:-

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- Investigations to exclude other causes
- **MDT :-**
  - Speech and language therapist
  - Occupational therapist
  - Educational psychologist
  - Community paediatrician
  - The mainstay of treatment is special education support. This is to help with language, social skills and communication skills.
  - Behavioural therapy which aims to reduce 'bad' behaviours and promote 'good' behaviours.

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Key INVX for unexplained developmental delay:-

= chromosomes
= fragile X
= Thyroid function tests
= Creatinine phosphokinase (CPK)
= Metabolic screen (urine amino acids , urine Glycosaminoglycans)
= X-Rays (hips/spine)
= Neuroimaging

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Key professionals involved in child management:-

= Paediatrician
= Physiotherapist
= Occupational Therapist
= Speech and language therapist
= Educational psychologist (if a child is likely to have special educational needs or benefit from specialized school or nursery placemet)
= Audiologist
= Ophthalmologist/ Orthoptist
= Neurologist
= Geneticist
= Gp
=Health Visitor
= Nursery/School Teacher

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Classic 'warning signs' of developmental delay

Fine motor/vision

○ Fixed squint at any age
○ Hand preference before **18 months** (usually not established until third year)
○ Not staring/no visual fixation or following **at 6 weeks**
○ Immature grip at **18 months**
○ Still casting at **18 months**

Speech/hearing

○ Failure to respond to sound at any age
○ Inability to understand simple commands at **18 months**
○ Not babbling at **12 months**
○ No spontaneous vocalisation at **18 months**
○ Unable to speak in short sentences at **2½ years**
○ Unable to understand speech at **2½ years**
○ Poor articulation making speech difficult to understand at **4 years**

Always remember to mention audiology referral if you notice delay in this area of development (and be aware of how hearing is tested in different age groups, see below).

Gross motor

○ Asymmetrical neonatal reflexes – up to **6 months**
○ Excessive head lag past **6 weeks**
○ Persistence of primitive reflexes (eg Moro/asymmetric tonic reflex at **6 months**)
○ Unable to sit or weight bear, absence of saving reactions at **12 months**
○ Not standing at **18 months**
  - In boys: think about Duchenne muscular dystrophy
  - If upper body gross motor development is normal (sitting with straight back, reaching for toys, normal head control): think about problems with hips or spine

Social/behavioural

○ Failure to smile: average age is 5 weeks, should be concerned if not smiling at **6 weeks**

History

1. **Personal information**
2. **Introduction, clarify aims**
3. **Present History:-**

   ACUTE:-

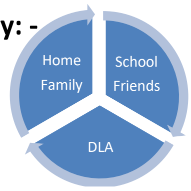
   - What is your main complain?
   - Would you please describe this .... for me ? (onset, course, duration, triggering, relief)

   ****** RELATED SYMPTOMS ******

4. **Other Systems Review:-**
   - ✓ **General:-** Does he look different? Any recurrent fevers?
   - ✓ **CNS:-** headache, change in behaviour, abnormal movements, weakness. Vision? Hearing?
   - ✓ **Neck:-** Thyroid :- prolonged N. Jaundice, constipation, sleepiness, Lumps, Bumps , wt loss, wt gain?
   - ✓ **Chest :-** recurrent cough, SOB, noisy sounds, change in colour.(interrupted feeds, sweating during feeds)
   - ✓ **CVS: -** exercise tolerance (interrupted feeds), sudden collapse, fainting attacks, change in colour.
   - ✓ **Abdomen :-**
     - ➢ **GIT:-** change of appetite, Nausea, vomiting, Reflux symptoms, Tummy pain, change in bowel habits (diarrhoea, constipation), Blood in stool, abdomen distension.
     - ➢ **Liver :-** yellowish discolouration of the eyes and skin, stool colour, bleeding
     - ➢ **Spleen :-** coughing of blood,
     - ➢ **Kidney :-** urine (stream, amount, colour) Dysurea, Loin pain, frequency
   - ✓ **MSK:-** restricted movement, Joint pain, any problems with walking?
   - ✓ **Skin:-** rash, nodules, discoloration.
   - ✓ **Heamatology:-** bleeding, pallor, recurrent infections

❖ **DD**

5. **Past History:-**
   - ❖ **Chronic Disease:-** When have you been diagnosed with..... For the first time? How was the first presentation? Have you ever been free of symptoms?
   - ❖ **Hospitalisation:-** how many hospital admissions per year? – did he require ICU admission?
   - ❖ **Invx:-** have you done any tests before ? (Blood, urine, stool, sputum, respiratory functions, HbA1c)
   - ❖ **Medication :-** Any regular medications ?dose, side effects, compliance , technique, who is giving it?.
   - ❖ **Surgery:-**
   - ❖ **Professionals involved so far:-** do you have regular FU? where do you FU? Who is your consultant?
   - ❖ **Complications:-** of this chronic disease
   - ❖ **Associations:-** with this chronic disease
6. **Perinatal History:-** is there anything you'd like to tell me about his birth history?
7. **Growth, Development & Puberty :-**
   - ○ **Growth :-** (**dietary history**) do you have his growth charts ? Following which centile? Wt gain? Wt loss?
   - ○ **Development:-** if a case with neurodisability or MSK you should discuss develop. In details :- - gross motor - fine motor -hearing & speech – social development **(Do you think that his movement and speech are normal in comparison to his peers ?)**
   - ○ **Puberty :-** (if child within puberty age) many children at the age of your child start to develop 2ndry sex characters like: -..... Did your child show this? Smoking, Alcohol, Drugs(if in puberty age)
8. **Vaccination & Allergy:-**
9. **Family History: -** + How many siblings he has? +Does anyone of them has the same problem? +Are they all healthy? +Are you and your husband relatives? +Any one of the family members has the same problem?
10. **Social History: -**



+ Who help in care?
+ EM: - do you know how to manage his emergencies like ..... ?
+ Social groups: - are you participating in any social groups?

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Psychological HX

Summarization with the mother

FINISHING:- ***what is your biggest worry?*** ***Are you concerned about any diagnosis in particular?*** ***Is there anything else that you would like to ask/tell me?*** ***Is there anything u haven't understood?***

Problem list:-

Summary: - Ahmed 12 years old child known case of ......, diagnosed when he was.... years old, coming complaining of a main complain of ....., and many other minor complains..... (Problem list)

Presentation

DD

INVX

Management :- don't forget admit or not ?

Ttt of acute medical condition

Management of chronic conditions

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